

SECOND EDITION

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*Principles &
Practice of*
**Pediatric
Plastic
Surgery**

Editors

Michael L. Bentz

Bruce S. Bauer

Ronald M. Zuker



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*For Kim, Gretchen, Alex, and Eric, who continue to make it hard to be at work,
because it is so fun being at home.*

M.L.B.



*To my wife, Sally, for unwavering support throughout my career, and acceptance
of her role as muse for my creative efforts, both surgical and artistic; to my family,
Erik and Mandy; and to my grandsons, Carson and Graham, who continue to provide
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B.S.B.



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R.M.Z.

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Preface

We are very pleased to present this second edition of *Principles and Practice of Pediatric Plastic Surgery*. As anyone who has edited a textbook will confirm, it is a labor of love and somewhat analogous to a maternity experience. We are particularly excited to present this updated and expanded edition for your daily use in the clinical practice of pediatric plastic surgery. The first edition was successful and well received, based on feedback we have received from practicing pediatric plastic surgeons around the globe. To improve many facets of these two volumes, we worked with our talented author teams and staff at Taylor & Francis/CRC Press, specifically expanding its content for your practical utilization. We have added seven new chapters that reflect the evolving contemporary head-to-toe practice of pediatric plastic surgery, encompassing surgical techniques and nonsurgical approaches to the children in your care. As our subspecialty has evolved, so has this textbook, reflecting truly state-of-the-art contributions from experts in the field. Because 8 years have passed since the first edition was published, we believe you will notice substantial changes that will help you care for infants, children, and adolescents.

In addition to content expansion and updating, this edition offers several additional features to help you and your patients. The book is now available as an e-book with full text and illustrations, featuring expanded operative video content to help you visualize intricate techniques. We have added new illustrations and expanded photographic examples, as well as colorizing key art from the previous edition. We have deliberately kept the book as a two-volume set to make it easier to transport and use as a reference, whether at your home, hospital, or while providing global health service.

With an expanding wealth of cutting-edge information in these pages, we have tried to review the topical problems and solutions for the more common anomalies, and we also present current innovations, offering solutions for defects for which previous solutions may have been accepted but were less than optimal. Perhaps of equal or greater significance is our effort to include the additional years of follow-up (up to 8 years) for cases that have been included in updated contributor chapters, placing emphasis on how important long-term follow-up is to our specialty. Our contributors represent an international cross-section of pediatric plastic surgeons and educators who have committed their best efforts to you on your patients' behalf.

We hope that this book will provide a source of stimulation to those new to our beloved field, while also serving as a source of continued motivation for those more experienced in our specialty. For laypersons reviewing this text, we hope you will feel supported through the challenging times of parenting a child with one of the problems that are treated by pediatric plastic surgeons, and know that the future is bright for your child and the children of others as they pursue their life dreams supported by their pediatric plastic surgeon.

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Although excellence in patient care drives our daily clinical effort, teaching and mentorship of junior colleagues, fellows, residents, and students are our most important educational responsibilities, and the ones that ultimately drive the future of patient care. I have been blessed to be surrounded by superb plastic surgery residents at the University of Wisconsin, Madison and the University of Pittsburgh; they have made the days (and many nights) truly worthwhile. That these junior colleagues are so far better than me in so many ways is a source of great pleasure.

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Michael L. Bentz



It is always difficult to decide who, in our personal and professional lives, has had a major influence on bringing us to this point, where some are specifically acknowledged, yet others, who may have been equally influential, are not specifically mentioned. So at the very outset, I apologize to anyone who I may have forgotten, and express my thanks for their help in whatever little accomplishments I have made.

Having grown up as the son and grandson of doctors, the choice of pursuing medicine as a career was no surprise. Both my parents, especially my mother, nurtured my artistic side and created an environment in which my sister, Laurie, and I had the tools to experiment and opportunities to reach for perfection in multiple manual crafts. Gradually, the idea of melding the science of medicine with my need to be manually creative led me to surgery.

Growing up with a father who was a child psychiatrist, I was also well aware of the potential psychological difficulties that children can face, how their feelings of self-worth are influenced by their parents and peers, and how varied forms of therapy, applied at appropriate points in their development, can profoundly affect their lives.

In the early months of medical school at Northwestern University I was introduced to the world of plastic surgery in a dazzling lecture by B. Herold Griffith, the Chief of Plastic Surgery. Approaching him afterward as an eager student, his warmth, enthusiasm, and constant desire to teach led to my decision to train in plastic surgery. He subsequently opened many doors for me: when I was still a medical student, he allowed me to become a regular visitor in the operating room at Children's Memorial Hospital, "assisting" Dr. Clarence Monroe. Later, as a resident, he connected me with my future Chief, mentor, and then partner, Dr. Desmond A. Kernahan. By the end of my training, I was committed to a career in pediatric plastic surgery.

Dr. Kernahan was a role model, with his meticulous attention to detail, innovation in surgical approach, and joy of surgery. Although he was generous in teaching me all of his tricks, he prompted me to seek my own surgical niche. This led not only to a concentrated interest in surgery of the ear, but also to a lasting relationship with our Pediatric Dermatology Service. My early work with Dr. Nancy Esterly, then Chief, launched me into the world of giant nevi, and her successors, Dr. Amy Paller and Dr. Tony Mancini, have continued to provide a never-ending supply of dermatologic challenges.

Throughout the inherent ups and downs of all academic careers, the joys of teaching residents and fellows has, I hope, provided as much inspiration for them as it has for me. I thank them all.

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their egos. I am honored to have this opportunity to work with both of them and am confident that our friendship will last throughout our lives.

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Finally, I must thank the many patients who have placed their trust in me and provided me with the impetus to strive for ever-better solutions to their varied problems. Organizations such as Nevus Outreach have furthered our understanding of specific defects and opened windows for comprehending the impact of that defect, not only on our patients, but also on their entire family. This brings me full circle to where I began and underscores why each of us has devoted our professional careers to this incredibly rewarding specialty of pediatric plastic surgery.

Bruce S. Bauer



As academic surgeons, our lives are so complex and intertwined with so many people and events that selecting milestones, career influences, and mentors can be extremely difficult. First, I would like to acknowledge my first Chief, Dr. W.K. Lindsay, who had the wisdom and foresight to recognize the value and potential of microsurgery in children. He gave me the opportunity and support to use this microsurgical approach to tackle complex clinical problems. I would also like to acknowledge my colleague in pediatric plastic surgery, Dr. H.G. Thomson, whose sensitive and caring manner with children and remarkable technical expertise served as my model in clinical practice.

The progress we made in microsurgery in children would not have been possible without the help of my microsurgical partner, Dr. R.T. Manktelow. His constant questioning of the status quo, searching for new solutions, and attention to detail stimulated me to the pursuit of excellence. Through the aid of a McLaughlan Traveling Fellowship from the University of Toronto, I was able to train in microsurgery in Japan under the leadership of Dr. Seichi Ohmori and Dr. Kiyonori Harii. I learned to operate using a microscope in the lab from the skilled and very patient microsurgeon, Dr. Shuhei Torii, with whom I remain close friends. Finally I learned to think outside the box and explore new concepts while training with my teacher, colleague, and friend, Dr. G. Ian Taylor. These are the mentors who supported me and allowed me to accomplish whatever small achievements I have made.

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My career has been punctuated by successes and challenges. However, no matter what, I could always depend on the support of my family as we crossed many bridges together. Although my heavy career commitment forced me to miss many of my children's milestones, we still had time to enjoy the best that life has to offer together. To my three sons and daughter-in-law, Jeremy, Melissa, Andrew, and Phil: I thank you for your patience and understanding. To my wife, Gail, of 42 years, who promised never to be upset when I was late: You have guided me, supported me, and assisted me at every turn. You are my true life partner. I thank you and will love you forever.

Ronald M. Zuker



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Part I



General Considerations in Pediatric Plastic Surgery

1

The Evolution of Pediatric Plastic Surgery

Bruce S. Bauer • Michael L. Bentz • Ronald M. Zuker



he field of pediatric plastic surgery offers endless opportunities for transforming the lives of children. As reconstructive surgeons, we can develop solutions to complex congenital conditions that previously represented insoluble problems. Although the challenges are great, the potential for positively improving the quality of life for these children is enormous. Few areas of medicine are as rewarding as pediatric plastic surgery. Our history is rich and our evolution fascinating.

HISTORICAL OVERVIEW

Historical Chinese documents from the Chin Dynasty (fourth century) clearly describe cleft lip surgery and the postoperative care required for success. In the sixteenth century, Ambroise Paré of France brought the repairs to life with his clear illustrations. Even Tagliacozzi, of rhinoplasty fame, entered the debate by emphasizing the need for muscle repair with cleft lip surgery. The masters of the past built the foundation of our current subspecialty.

Congenital nevocellular nevi are referred to in the Old Testament when Jacob selected his spotted herd.¹ After Jacob's wife Rachel gave birth to Joseph, he asked her father, Laban, to let him go. For payment for all his work over the past 20 years, he asked Laban for the spotted sheep and goats. To increase his compensation, Jacob created a striped environment from peeled branches, where the nonspotted animals would drink and mate. They bore spotted offspring that then would belong to Jacob. This substantiated the view at the time that visual stimuli can affect intrauterine development. The spots on the animals were thought to be a result of the herd's environment.

In the Middle Ages, nevi and birthmarks were blamed on the behavior of pregnant women, who were thought to be "consorting with the devil." Currently, the exact cause is not known,

be it environmental, genetic, or simply a random mutation. Although superstitions still provide explanations for congenital deformities in some parts of the world, rapid advances in the allied sciences, particularly genetics, move us toward more concrete explanations of many defects. Hopefully, these advances will lead to reduced occurrences of some deformities and provide the information necessary to modify our surgical techniques or sequences of procedures to improve the long-term outcome of our reconstructions. We are part of an ever-evolving specialty.

Pediatric plastic surgery had its origins as a specialty in 1982 with the founding of the American Association of Pediatric Plastic Surgeons. The goal of this society was to provide pediatric plastic surgeons with educational opportunities and continuity and camaraderie within the plastic surgery community. The move to raise awareness of this “new subspecialty” within the field of plastic surgery was reinforced by the subsequent formation of a Plastic Surgery Section of the American Academy of Pediatrics in 1987, which emphasized that plastic surgeons who devoted a substantial part of their practices to children and qualified to be a Fellow in the American Academy of Pediatrics (FAAP) were separate and distinct from their general pediatric surgery colleagues. As this subspecialization developed elsewhere in the world, the subspecialty of pediatric plastic surgery was born.

Pediatric plastic surgeons have become an integral part of the health care of children. As in other areas of surgery, when plastic surgeons began to focus on children and became more subspecialized, the widening recognition of their services and abilities among their colleagues and among grateful parents stimulated referrals, thereby expanding the scope of treatment. As more pediatric plastic surgeons played a role in other pediatric subspecialty clinics (such as those for dermatologic or vascular lesions), they encountered an increasing variety of difficult problems and had to find new ways to manage them. As more patients with similar problems were treated, the scope of the field widened, and a new niche was established within the field, making pediatric plastic surgeons indispensable team members at all children’s hospitals and at hospitals treating large pediatric populations.

Early pediatric plastic surgery focused on treating cleft lip and palate, microtia, and congenital hand anomalies. It quickly embraced craniofacial surgery, which led to an impressive expansion in types of treatments and surgical procedures available. Contemporary pediatric plastic surgeons have witnessed a rapid extension of treatment to regions other than the cranium, including the trunk and the extremities. At these sites, our discipline has made significant contributions, independently and with other pediatric surgery specialists and subspecialists, to the care of children with problems not commonly associated with traditional pediatric plastic surgery.

Initially, plastic surgery procedures performed in children were the same as those used in adults. Over the past 30 years, however, pediatric plastic surgery has developed its own unique procedures, and the specialty has blossomed into a vibrant area of practice. New techniques have arisen, and new problems have been solved. Advances in surgical theory, technique, and technology now offer children with deformities (and their parents) the promise of a wonderfully ordinary childhood and an opportunity to enter the adult world without the obstacles that previously marked them as outsiders.

More than just plastic surgery on little people, pediatric plastic surgery takes a typical three-dimensional reconstruction and adds a fourth dimension—time. Surgical decisions require consideration of both the effect of the surgery on growing structures and how the results will be changed by subsequent growth of the head and neck or body region. Long-term follow-up has now allowed techniques to be further refined to minimize the impact of surgery.

Although the training of plastic surgeons and general surgeons has been comingled historically, more recently the curriculum has diverged for many plastic surgeons through residency training models, the scope of practice, and independent division versus departmental adminis-

trative status. To a list of long-established craniofacial fellowships, a slowly increasing number of pediatric plastic surgery fellowships is being added to train young surgeons in the full spectrum of pediatric plastic surgery. These neophytes are not only completing their plastic surgery education but also setting the stage for a career in which pediatric plastic surgery plays a major role.

ADVANCES IN PEDIATRIC PLASTIC SURGERY

Great progress in pediatric plastic surgery has been made through the years. The advances discussed here have been essential for restoring form and function for thousands of young patients around the world.

Advances in Cleft Lip and Palate

A greater understanding of cleft anatomy and the effects of surgery on the growth process has led to significant progress in the treatment of cleft lip and palate, which in turn has led to improved outcomes and a reduced need for lip revision surgery. Presurgical orthopedics has assisted surgeons in the primary lip repair, also leading to improved outcomes. Advances in nasoalveolar molding and primary nasal repair have reduced severe nasal deformities. Cleft lip repair continues to evolve. Mohler² and Fisher³ have developed new techniques, striving toward even better results. Palatoplasty is now carried out earlier with better speech outcomes. New palatal procedures have evolved and are now well established, such as Furlow's double-opposing Z-plasty repair,⁴ which is one of the more significant palatoplasty techniques to be described in the past 75 years.

Progress has also been made in bone grafting of nasoalveolar clefts, the application of distraction osteogenesis for early treatment of select cases of severe maxillary hypoplasia, and early treatment of mandibular hypoplasia in patients with Pierre Robin sequence and in patients with syndromes affecting overall development of the facial skeleton with compromise to the airway.

Advances in Craniofacial Surgery

Craniofacial surgery is one of the areas in which some of the most impressive advances have been made in plastic surgery. Today, major improvements in operative technique allow surgeons to operate on infants with decreased blood loss, fewer complications, and increased safety. Other improvements in this area include a better understanding of the long-term effects of the various syndromic and nonsyndromic craniosynostoses on surgical outcome, the use of absorbable plate fixation, and an ever-increasing number of bone substitute materials.

In addition, pediatric craniofacial surgery has seen an increase in the use of distraction osteogenesis to treat hemifacial microsomia, mandibular hypoplasia, severe maxillary hypoplasia with airway and orbital compromise in Apert syndrome, and other severe craniosynostosis syndromes. There have been improvements in the treatment of hypertelorism and in our understanding of how growth affects the surgery. This has changed the timing of surgery to perhaps minimize the amount of relapse and limit the number of surgical interventions. Better imaging techniques and computer-based modeling and planning have had an enormous impact on the accuracy of craniofacial reconstructions and continue to evolve and facilitate improved outcomes.

Another major step forward has been an ever-expanding knowledge of the genetic basis of craniofacial syndromes, which alone may lead to a gene-based reclassification of syndromes and possible later application of gene therapy. On the technical side, the increased use of absorbable plate fixation and absorbable distraction devices has helped to limit the number of surgical procedures for these children.

Advances in Skin and Soft Tissue Reconstruction

Advances in tissue expansion for treating congenital giant nevi and large soft tissue and skin deficits of congenital and acquired origin represent another important milestone. Tissue expansion has served as an adjunct in select microvascular and craniofacial reconstructions. There are future prospects for the use of skin substitutes for resurfacing burns and congenital nevi.

New approaches to the timing of reconstruction have been instrumental in improving outcomes. Early reconstruction for complex problems such as congenital nevi and a comparison of matched cases with various reconstructive techniques lead to better-designed procedures that withstand the test of time and growth, thereby limiting the number of later reconstructive procedures required. Although early and sometimes complex surgery places a greater burden on the parents and family, an affected child may be young enough to be spared at least some of the physical pain and much of the psychological trauma of later surgery.

Advances in the Classification and Treatment of Vascular Lesions and Anomalies

A greater understanding of the biology of vascular lesions and improved classification methods have led to early medical treatment of hemangiomas, followed where needed by appropriately timed surgery. Interventional radiology for treating vascular malformations and free tissue transfer in the reconstruction of complex arteriovenous malformations is possible now, along with laser treatment of capillary vascular malformations and select hemangiomas. In addition, nonsurgical treatments are available for vascular anomalies, including sclerotherapy and embolization, either alone or as an adjunct to surgery. Therapies affecting the growth of vascular lesions at the cellular level are anticipated.

Advances in the Treatment of Trunk Anomalies

The use of “commonplace techniques” has revolutionized reconstruction of the thoracic and abdominal wall for children with the difficult omphaloceles, gastroschisis, pentalogy of Cantrell, and ectopia cordis anomalies. Patients requiring open correction of pectus excavatum are still well served by Ravitch-based techniques, relying on the routine plastic surgery principles of flap mobilization, resection of anomalous anatomy, and fixation, leading to mutually important functional and aesthetic reconstructions. Intraabdominally, microsurgical anastomosis of the hepatic artery has been shown to increase vascular patency in split-liver transplants, improving survival. Standard microvascular techniques have been successfully applied to treat patients with renal artery stenosis. The cross-pollination of these techniques would not have occurred in the absence of our daily responsibilities as the surgical cavalry for our colleagues and patients.

The Back

The back has proved to be a significant source of patient care problems and solutions. Day-of-birth flap closure of meningomyeloceles, later flap coverage of exposed back hardware, intrathoracic muscle flap transposition for bronchopleural cutaneous fistulas, and empyema dead space fill have extended survival rates for patients with historically difficult wounds.

The Breast

As improved breast procedures have been developed for treating adult patients, similar progress has occurred for pediatric and adolescent patients. Young women no longer need to live with significant breast asymmetry and hypoplasia, which directly affect socialization and healthy par-

ticipation in physical activities, such as gym class, team sports, and recreational athletic participation. More and more infants survive extended stays in the neonatal intensive care unit. The unanticipated breast size and shape consequences of early cardiac and thoracic interventions can be corrected or temporized before additional unfavorable effects occur.

Advances in Microsurgery

Replantation of amputated parts in children preserves function that far exceeds what is expected in adults. Virtually any amputated part of a child that can be replanted should be replanted. Free tissue transfer offers solutions to enhance form and function for congenital anomalies of the head and neck. These include bony and contour restoration for hemifacial microsomia and reanimation techniques with functioning muscle transplants for facial paralysis. For congenital deficiencies of the hand, procedures such as toe-to-hand transfers are performed for patients with symbrachydactyly. For children with cancer, pediatric plastic surgeons and pediatric microsurgeons are integral parts of the team approach. With improved chemotherapy, the surgical excision of sarcomas can lead to high cure rates. However, the defects of surgery, whether bony or soft tissue, require reconstruction for optimal function and quality of life. Pediatric microsurgeons can facilitate the extirpative cancer surgery by providing an acceptable functional and aesthetic reconstruction. This applies not only to soft tissue reconstruction but also to bony reconstructions and the newer joint reconstructions. Secure coverage, including the bringing in of well-vascularized tissue for allograft coverage or prosthetic joint support, vascularized bone for extensive bony defects, and even vascularized growth plate transfers, provides opportunities for surgical success in pediatric sarcoma management. Pediatric microsurgeons work with smaller but generally healthy, normal vessels and healthy children, who recover rapidly.

Advances in Other Areas

Pediatric plastic surgery has made strides in several additional areas, some involving new techniques and others relying on procedures adapted from other facets of reconstructive surgery, such as the following:

- *Ear reconstruction:* Reconstruction of the external ear for microtia and acquired ear deformities has improved, with better aesthetic outcomes, a reduced number of surgeries, and a reduced risk of complications.
- *Extremity surgery:* The treatment of congenital hand deformities; the treatment of obstetric brachial plexus injury; microvascular surgery for complex, posttraumatic composite reconstruction; and the use of distraction for congenital hypoplasia of the digits have advanced greatly in recent decades.
- *Separation of conjoined twins:* As integral members of the separation team, pediatric plastic surgeons can provide appropriate reconstruction and cover to facilitate successful separation surgery for conjoined twins.

Although significant progress has been made, much work remains to be done. Our understanding of vascular anomalies has grown substantially, but our treatment options have not expanded proportionally. We have treated these lesions in an improved anatomic and physiologic fashion, relying on a multidisciplinary effort; however, cell-specific intervention is essential.

Abdominal wall transplantation, although useful at present, should lead to a tissue-engineered solution, followed by a stem cell-mediated solution, eliminating the long-term consequences of immunosuppression in children. Significant progress has been made. However, much more work is necessary. Specifically, our treatment options for vascular anomalies fall short, despite

improvements. Perhaps a cell-specific intervention is possible. Similarly, as the era of vascularized composite allotransplantation in children nears, a tissue-engineered, stem cell-mediated solution becomes possible. Finally, a clearer understanding of fetal wound healing, tocolytic therapy, and the successful evolution of intrauterine preterm surgery will allow us to harness the vast potential of scarless healing. The challenges are endless!

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Global Health in Pediatric Plastic Surgery

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Surgical disease has been estimated to account for 11% of the world's disability-adjusted life-years (DALYs), or years lost to death and disability.¹ An estimated 14 million DALYs are related to congenital anomalies, and 62 million DALYs are caused by traumatic injuries.² In addition, road traffic injuries are a leading cause of death among individuals 10 to 19 years of age, and burn injuries necessitate hospitalization for an estimated 500,000 children each year.^{3,4}

Despite these staggering numbers, approximately 2 to 3 billion people do not have access to basic surgical care.⁵ Furthermore, there is a significant disparity between the availability of surgical services in resource-rich countries and those of resource-poor countries. A review of surgical volume for resource-poor countries, which account for 35% of the world's population, showed that these countries were responsible for only 3.5% of all surgical procedures performed globally.⁶

Surgical outreach has historically been omitted from global health agendas, possibly because the focus has been on communicable diseases.^{7,8} Interestingly, noncommunicable diseases have been shown in several instances to account for a larger burden of disability than communicable diseases.⁹ An increase in the number of disabilities and deaths on the African continent, for example, is related to surgical rather than infectious disease; historically, however, more attention has been given to parasitic, bacterial, and viral diseases.¹⁰

Considering the great volume of surgical disease attributed to congenital, burn, and traumatic bony and soft tissue injuries, plastic surgeons have a significant role to play in global health. However, there is a significant shortage of plastic surgeons in many countries, including Zambia (1 plastic surgeon per 10 million people). Other more populous nations, such as China and India,

have an estimated 3000 qualified plastic surgeons.¹¹ Yet as in wealthy countries, many surgeons in these countries are drawn to urban lifestyles and cosmetic surgery practices.¹¹ Predictably, surgery is often only available to the wealthy, which exacerbates the problem.⁸

Thus there is a tremendous need for interested plastic surgery volunteers to contribute to treating surgical disease in the short term and creating sustainable models for ongoing prevention and care of patients in the long term. Many of the examples in this chapter will focus on the history and evolving models for cleft lip and palate surgical outreach, because this has been the focus of the majority of research in the plastic surgery and global health literature. Nonetheless, pediatric plastic surgeons have an integral role to play in the various disease processes regularly encountered in practice.

GLOBAL HEALTH MODELS

Global health programs have often been distinguished as representing a vertical or horizontal model of health care delivery. *Vertical models* involve the delivery of health services from donors and volunteers without integration into local health systems. *Horizontal models*, on the other hand, involve the provision of health care services through publicly financed health systems and local health care providers.

The concept of horizontal health care delivery was established and advocated for in the World Health Organization (WHO)/United Nations Children's Fund (UNICEF) declaration of Alma Ata in 1978.¹² The declaration of Alma Ata was the first international declaration emphasizing the importance of primary health care for all citizens of all nations. There was a strong emphasis on primary care, the importance of the state in providing health services, and access to and equality of services for all people. Various failures of horizontal initiatives, such as the inability to contain malaria in India in the 1980s, may have encouraged a shift toward predominantly vertical strategies for the delivery of health interventions to underserved populations.¹³ Vertical programs often have more immediate results than horizontal programs, because they do not require working through local government systems and may therefore be more attractive to benefactors.

Although the dichotomy between vertical and horizontal health delivery strategies is often made, a systematic review of global health programs identified that the programs often employ a combination of strategies, some of which are integrated into the local infrastructure and others which are not.¹⁴ Multiple considerations must be addressed in any outreach effort, ranging from financing and revenue generation for a project to administrative planning, priority setting, resource allocation, health service delivery, program monitoring, and patient education.¹⁵ Some of these considerations may not be manageable in a local context given limited resources, whereas other strategies might be entirely possible with local personnel and infrastructure, leading to hybrid programs with a mix of international and local contributions.

In the context of global surgery, a few models have been described that involve varying dependence on both international volunteers and local personnel and resources. Although numerous organizations exist that provide charitable work to underserved areas, it is beyond the scope of this chapter to list them all or define what model these organizations represent. This would be challenging, because the organizational structure and health care delivery model of an organization may evolve over time. In addition, there are often vastly different opinions about what type of model a program represents based on what features of a program are emphasized. Operation Smile, for example, has been described as being organized around short-term medical missions, and it has also been described as promoting sustainability and the development of comprehensive cleft centers.^{16,17}

Rather than categorize all the major plastic surgery outreach programs, we will outline a few basic strategies that describe how a global surgery program may interface with an underserved area. We will also select a few examples of historical and current programs that exemplify certain features of these models, although one could argue that they also share features of other global health models.

Short-Term Surgical Case Performance Model

The short-term model involves surgical teams that make short visits to areas of need and perform a high volume of surgical procedures. The positive aspects of this model are that a large number of children can have surgery performed. The limitations of this model are that these programs do not necessarily establish local strategies for creating sustainable change, such as training local health care providers to perform surgery. Trips are usually brief, lasting a couple of weeks. Although ReSurge International has since changed its approach to have more of a focus on local capacity building, early efforts in the organization's history toward making periodic trips to underserved areas and performing cleft lip and palate, hand, and burn surgeries serve as examples of the short-term surgical trip.¹⁸

Another example is disaster relief in a location afflicted by war or natural disaster. A multidisciplinary team aboard a U.S. Navy ship provided comprehensive services in craniomaxillofacial trauma reconstruction after the 2010 Haiti earthquake.¹⁹ The ship was stationed near Haiti for approximately 2 months, which is longer than the duration of most short-term trips, during which 93 craniofacial reconstructions were performed. More recently, ReSurge has demonstrated that well established surgical teams can provide both long-term care in a region and disaster relief in scenarios such as the Nepal 2015 earthquake.²⁰

Hybrid Model

The hybrid model represents a cooperative partnership between the global health program and local plastic surgeons. The strength of this model is that it strives to deliver sustainable plastic surgery care to the location of interest. The effects of the outreach last well beyond the time frame of the outreach effort because of integration with local surgeons, who receive training to perform subspecialized surgery. Although fewer cases are performed in the immediate setting of a surgical trip, a multiplier effect happens, because local surgeons become trained and empowered to offer services to the community throughout the year. ReSurge similarly serves as an example of the hybrid model, because it has placed increasing emphasis on education and training of local health care providers to foster local plastic surgeon autonomy.¹⁸

SmileTrain also heavily emphasizes the empowerment and training of local health care providers by offering funding and educational resources to existing facilities that have the basic skills and tools but not the resources to expand their cleft outreach efforts.¹⁸ Their educational programs have included symposia, workshops, fellowships, and the use of virtual surgery software and three-dimensional animated videos to help facilitate the training of health care providers worldwide.²¹

The Barsky unit of Vietnam serves as a historical example of how a hybrid model can eventually transition to ownership of an outreach program by local physicians. In the wake of the Vietnam War, Children's Medical Relief International was founded and established a hospital for pediatric surgery in Saigon. Operations were initially performed by international volunteers, who also provided training to Vietnamese surgeons.²² The surgery center, also known as the Barsky unit, was established in 1969 and by the mid-1970s was almost entirely operated by Vietnamese surgeons.¹⁸

Extended Residence Model

In the extended residence model, the plastic surgeon relocates to an underserved area to develop a center of excellence in a resource-poor area. This model helps further the goal of sustainability by integrating the plastic surgeon into the hospital administration and community. As a result, there is ample opportunity for information exchange, continuity of care, and patient follow-up. A limitation of this model is that the plastic surgeon must be willing to leave behind or relocate family for an extended period of time.

Dr. Samuel Noordhoff's extended stay in Taiwan exemplifies this model. He initially moved to Taiwan in 1960 and worked as hospital superintendent at the Mackay Memorial Hospital for 16 years. He then became the chairman of the Department of Plastic Surgery at Chang Gung Memorial Hospital (CGMH) and established the Noordhoff Craniofacial Foundation (NCF), which helped support the development and eventual independence of the Chang Gung Craniofacial Center.²³ The craniofacial center now sends surgical teams to other countries in close collaboration with the NCF to help provide humanitarian care. The center also hosts international visitors and fellows, with the goal of cultivating autonomous care by local health professionals in other underserved regions.²⁴

OUTCOMES AND GLOBAL HEALTH IMPACT

Financial Impact

The United States has been estimated to be responsible for 6000 short-term medical and surgical trips to resource-poor countries each year through a variety of organizations at an estimated total cost of \$250 million. Medical trips are estimated to cost approximately \$3 per patient in contrast to the surgical cost of \$700, which accounts for equipment expenses.²⁵ Although these data do not stratify surgical cost by the type and length of procedure performed, they raise the question of whether surgical outreach trips are cost-effective.

Multiple strategies for determining the cost-effectiveness of plastic surgery outreach missions depend on the concept of the DALY. The number of DALYs related to a disease process refers to the number of years of life lost and the number of years lived disabled as a result of the disease.²⁶ The calculation of cost of an intervention per DALY averted depends on several variables, including costs of the intervention, life expectancy of a population, and possibly on the age of the patient if age weighting is used. With age weighting, greater value is assigned to the middle years of life, when the patient's social and economic importance is assumed to be greater.²⁷

An additional variable that goes into the DALY calculation is the disability weight for a disease process. Disability weights are provided by WHO and estimate the severity of a disease on a scale from 0 (healthy) to 1 (dead).²⁸ For example, the disability weight for untreated tuberculosis in the 0 to 4 age range is 0.294 and for a radius or ulna fracture is 0.180, compared with the disability weight of 0.098 and 0.231 for cleft lip and palate, respectively.²⁹

Once the cost per DALY averted is calculated, various strategies can be used to determine whether a given intervention is cost-effective. One approach is to use static costs as a reference for whether an intervention makes financial sense. In 1993 the World Bank published a range of prices that were considered cost-effective.³⁰ An intervention costing \$25 per DALY averted would be highly cost-effective, and an intervention costing \$50 to \$150 per DALY averted would be cost-effective. Any intervention can be weighed against these figures to estimate its cost-effectiveness.

Another strategy suggested by WHO has been to compare the cost per DALY averted with the gross domestic product (GDP) per capita in a country as a reference indicator of whether the cost to add a year of healthy life to an individual enhances the GDP.³¹ A cost-effective

intervention would be at least one time the GDP per capita, and it may be up to three times the GDP per capita, considering the potential of a healthy individual to contribute to and stimulate the economy. For example, Vietnam's GDP per capita is \$1051, and thus a cost of \$68 per DALY averted for cleft lip and palate repair is highly cost-effective.³²

Given the fact that surgical interventions have not been prioritized at the international level in favor of other public health interventions aimed at infectious disease, another strategy is to compare the cost per DALY averted for a surgical intervention directly to the cost of other well-accepted public health interventions, such as vaccination or tuberculosis screening.³² Comparisons reveal that surgical reconstructions such as cleft lip and palate repair are cheaper than sanitation efforts for diarrheal disease and programs directed at prevention of maternal-fetal HIV transmission.

Identifying the cost per DALY averted is clearly important regardless of the strategy used to define cost-effectiveness. A general range of costs for common surgical procedures performed in low- and middle-income countries has been described as \$70 to \$230 per DALY averted.³³ Further data are available for the cost per DALY averted for cleft lip and palate surgery specifically. A review of expenditures for eight Operation Smile missions in 2008 showed that primary cleft lip and palate repair is cost-effective.³⁴ The analysis in this study focused on equipment and travel expenses and did not include the costs to the patient for travel or to the local hospital for accommodating the team. The authors' calculation of cost per DALY averted incorporated the hypothesis that cleft lip and palate cause lifelong disability. The cost per DALY averted ranged from \$7.36 to \$96.04 (average \$33.94), and when these costs were compared with other public health measures, it was clear that the expenditure per year of disability averted is cost-effective.

A study examining the cost-effectiveness of cleft lip and palate repair in Vietnam not only accounted for supply costs and the costs to the outreach organization, but also the costs for patient transportation, food, lodging, and hospital costs.³² The cost estimated per DALY averted for cleft lip and palate surgery in Vietnam ranged from \$56 to \$97, depending on whether age weighting is used and whether the time spent by visiting Korean surgeons is considered vacation time or lost salary. This cost falls in the range of other public health programs, including HIV/AIDS education, and is more cost-effective than many other public health interventions aimed at family planning and tuberculosis management. Further studies have confirmed that cleft surgery trips are highly cost-effective, with an estimated cost of \$72 to \$134 per DALY averted in outreach efforts by SmileTrain.³⁵

Although studies have assessed the cost-effectiveness of global surgery services, the data on quality of care delivered by both visiting and local surgeons have not been adequately studied. Data are scarce on early and late surgical complications. Residual disability related to an absence of speech therapy for cleft palate patients and occupational therapists for patients with hand and burn injuries has not been well described.³² In addition, data on patients' age of intervention, surgeries they underwent before plastic surgery trips, and related costs and consequences of these interventions deserve further attention.¹⁶

Academic Impact

The potential academic and training benefits of global health trips are to health care providers from both volunteer and recipient countries (Fig. 2-1). Plastic surgery resident volunteers may wish to improve their cultural competency and understanding of other health care models, appreciate the impact of global outreach projects, and have the experience of operating in a setting with limited resources. Local providers in resource-poor areas may want to develop new surgical subspecialty skills, acquire new technologies and resources, and develop collaborations and opportunities for advanced subspecialty training.



Fig. 2-1 Educational exchange is an important aspect of global health initiatives and may occur in many forms. **A**, Nicaraguan and University of Wisconsin (UW) plastic surgery residents work together under close supervision to harvest a rib graft for ear reconstruction. Residents learn by operating with both Nicaraguan and UW faculty and from each other. **B**, A UW faculty member takes a Nicaraguan resident through a cleft procedure with other trainees assisting.

There is increasing interest among surgical residents in the United States in having exposure to surgical volunteerism during training.³⁶ A survey of 55 North American surgery programs revealed that 7 (12%) have formal international rotations for residents, 11(20%) have health-related partnerships, and 34 (62%) have informal programs.³⁶ Experiences among these programs revealed the importance of finding strategies to balance the experience of North American resident volunteers and local trainees, as well as the importance of adapting the goals of a surgical trip to the specific needs of the local community.

A review of the training experiences of plastic surgery resident volunteers specifically reveals that international volunteerism contributes significantly to their education. Survey data for 19 plastic surgery residents who participated in a surgical outreach trip showed that all residents felt an improved sense of cultural competence, enhanced awareness of the nuances of planning and implementing global surgical outreach, and an improved knowledge of how to correct cleft lip and palate deformities.³⁷ Most respondents also felt that their surgical skills were improved, confidence was gained, and the experience provided the foundation for future outreach to underserved populations. A survey of American plastic surgery residencies offering global health experiences found that 96% of programs endorse these opportunities, particularly because they provide exposure to other training systems and enhance surgical education.³⁸ Programs that were unable to offer global health experiences cited the lack of case accreditation and funding as frequent barriers to these cross-cultural and surgical opportunities. These barriers warrant further attention to improve resident access to global surgery opportunities.

Although there are abundant data demonstrating a benefit of global surgical trips to resident volunteers, no studies were identified that rigorously assess the educational value to local health care providers. The potential value may be not only to local physicians but also to surgical technicians, who can perform simple procedures in areas with a physician shortage. Training nonphysicians to perform specialized surgical procedures is a controversial matter. Some would argue that this hinders the development of high-quality health care among surgical subspecialties and leads to fragmentation of services, with poor monitoring and surveillance. On the other

hand, it may be argued that certain simple procedures, such as splinting a fracture or skin grafting a wound, could be performed by individuals with minimal medical training. For example, studies have shown that midwives and technicians can be trained to provide basic surgical interventions to help expand access to emergency obstetric care.³⁹ In addition, nonphysicians who perform surgical interventions may be more likely to provide long-term care in rural and underserved areas.⁴⁰ The potential training benefits of global surgical trips may extend further to local team members providing speech therapy, orthodontic, and other important ancillary services.

In addition to the direct training benefits of global surgery experiences, the pairing of academic programs from volunteer countries with local communities may enable novel research on the needs of underserved areas and methods of delivering cost-effective interventions. Such research may improve the delivery of effective, sustainable care and impact global health policy.⁴¹ In addition, genetic and epidemiologic studies in underserved areas may foster new strategies for disease prevention.⁴²

A North American academic general surgery program demonstrated that time spent in a resource-poor setting by residents during their research years could help improve the quality of global health outreach.⁴³ Resident volunteers sent to Malawi under the supervision of university and local surgeons assessed the training needs of local providers in Malawi and were able to use their needs assessment to improve the curriculum for training local health providers in surgical specialty care. Resident research was also useful for better defining the surgical needs of the local population and showed that WHO estimates of traumatic injuries overlooked patients situated remotely from the hospital. Patients who died as a result of poor access to immediate trauma care were not incorporated into trauma registries. The role of residents in conducting research used to improve the quality and content of training received by local health care providers demonstrates how academic partnerships may benefit local communities.

Sustainability

Survey results from short-term medical and subspecialty surgical trips suggest that 80% consider themselves to have a collaborative relationship with local health care systems, and 100% have a strategy for resumption of care by another provider in case they are no longer available.²⁵ Although collaboration with local providers is important, developing a sustainable project for an underserved community requires that the project meet several criteria, as outlined by WHO⁴⁴:

- The project is effective.
- The project has longevity.
- The project has high treatment coverage.
- The project is integrated into existing health care services.
- The project is owned by the community.
- The project relies on resources from the community and the government.

Previous concerns have been raised that plastic surgery global outreach trips often focus too much on the number of patients they treat rather than the important role of training local plastic surgeons to provide affordable, quality care.⁴⁵ Furthermore, failure to integrate volunteer services into local systems may result in lack of follow-up on patient complications after a volunteer group has left an area.⁴⁶

A study involving group discussions with medical officers in African countries revealed mixed opinions about international volunteers.⁴⁷ Foreign volunteers were valued for their service to remote areas, their ability to provide organization and motivation to local providers, and their skills in educating and training personnel. However, there was also a perception that surgical trip participants were inexperienced, limited by cultural and language barriers, and did not help strengthen the public health system, therefore limiting their long-term impact.

The importance of transitioning ownership of health systems, including surgical interventions, to local teams cannot be overstated. There are multiple potential reasons, however, why outreach programs have not been successful at integrating their volunteer efforts at the local level. In many scenarios, volunteer groups have found that available staff, training, and resources are inadequate to meet the needs of a community.⁴⁸ Furthermore, donors may be more willing to fund quick, short-term interventions, because long-term changes in another country's governance or resources may be unpredictable. Another reason that integration may be challenging is that many parts of the world have questionable government legitimacy and rebel groups who would see outreach efforts through the government as undermining their cause.⁴⁹

To address some of the limitations to immediate integration of a global health outreach program into underserved areas, the concept of a diagonal model has been proposed that takes advantage of foreign resources while maintaining a vision for long-term integration and capacity building.⁵⁰ In this model, foreign volunteers, money, equipment, research ideas, and surgical interventions are welcomed into a resource-poor area, as long as strategies are found to link these resources to sustainable systems that will be widely accessible. In many ways, the diagonal model describes what many outreach programs have already been trying to do.

Literature published on outreach efforts by Operation Smile provides an example of how over time there has been an increasing commitment to not only provide immediate interventions but also to build local infrastructure for sustainable change in delivery of pediatric plastic surgical care.⁴² Several realizations led to increasing interest in local capacity building. Volunteers were limited, and many children would be left untreated without the expansion of local resources and providers. Mission trips that failed to incorporate local providers would potentially undermine and alienate them. A whole team of personnel, including speech therapists, orthodontists, and pediatricians, would be necessary to provide comprehensive care. Systems would need to be in place to ensure the availability of staff trained in life support to maximize patient safety perioperatively.

The importance of a multidisciplinary team in ensuring a sustainable global outreach program, however, goes beyond the roles of physicians in ensuring a safe perioperative experience for the patient. Social workers and psychologists may play an important role in discussing the implications of a pediatric disease process like cleft lip and palate while allaying fears and providing education to families.⁵¹ A multidisciplinary team may also be able to better address malnourishment in children, which is a frequent finding in resource-poor countries that may contribute to complications with wound healing.⁵²

Furthermore, multidisciplinary teams may help improve patient follow-up, which historically has been poor for patients receiving cleft lip and palate surgery during global surgery trips. A 6- to 9-month follow-up for a global cleft surgery endeavor that performed 4100 operations in 2007 was only 36.7% at selected sites.⁵³ A 1-year follow-up was even lower, at 19.9%, and many pediatric plastic surgeons would argue that the 1-year time point is too early to make an accurate assessment of speech outcomes, facial growth, and aesthetics. Therefore, an outreach team with a nurse coordinator or social worker who can facilitate follow-up or find ways to provide patient transportation would be helpful.

Given the importance of developing a multidisciplinary team with local roots, the promotion of surgical specialty hospitals may be one of the most effective ways to deliver quality surgical care safely and affordably.¹⁶ Research in the United States has shown that surgical outcomes are related not only to surgeon volume but also to hospital volume.⁵⁴ The development of centers of excellence for global pediatric plastic surgery that are staffed and led by local plastic surgeons may help ensure the enduring availability of surgical services to populations in need. Whatever the strategy, there must be a carefully considered plan for the long term in any plastic surgery mission as part of the ethics of global health.⁵⁵

Independence

Evidence exists that pediatric plastic surgery initiatives can take root in underserved areas and become autonomously functioning entities. The common theme to stories of locally established centers becoming independent is a strong, long-term commitment to the community on the part of international collaborators with a significant investment in the training, financing, and resourcing of local plastic surgeons.

A commitment by a Korean initiative called Smile for Children to underserved communities in Vietnam resulted in a partnership in which Vietnamese staff had significant autonomy.³² Initially, surgical trips by Smile for Children were led by a senior Korean surgeon and Korean surgical residents. An emphasis during surgical trips was to include Vietnamese surgeons on the operative team to help them learn operative techniques in cleft lip and palate surgery. Vietnamese surgeons were also sent to Korea for intensive surgical training. The result of this approach was that the Vietnamese group continues to operate year-round and only relies on visits from the Korean group for more complicated cases and ongoing learning opportunities.

The NCF, in partnership with CGMH, has also helped to create self-sufficient cleft programs at the local level and to describe a series of steps for achieving this goal. Ironically enough, the CGMH Cleft and Craniofacial Center (CCC) itself was a product of slow, gradual development. When the CCC began, there was only one plastic surgeon, and there was a lack of trained personnel, funding, and government interest, because the public health focus was on treating infectious disease.⁵⁶ Currently the CCC works in conjunction with the NCF to provide cleft training and outreach to underserved areas and to promote their independence. A key aspect in the development of a multidisciplinary center was the selection of motivated people.

The NCF and CGMH have described a series of developmental stages for the emergence of local cleft programs²⁴:

- Stage 1 involves identifying a country or institution to become involved with the provision of services and selection of key physicians and personnel for further training.
- Stage 2 involves providing further training to key physicians and personnel. CGMH reports training 92 “seed” physicians and personnel from 12 countries from 2000 to 2008.
- Stage 3 involves establishing a local cleft center with details decided about facilities, staffing, and funding. At this stage it is critical that local health authorities and providers are willing to support the cleft center logistically and financially to increase the likelihood of it becoming independent of foreign support.
- Stage 4 involves establishing a local cleft foundation that is financially capable of supporting not only itself but also of delivering aid and training to other underserved areas.

The NCF and CGMH report that they have helped to establish a local cleft foundation in the Philippines and cleft centers in Cambodia, China, and Indonesia. Although the global health literature often describes outreach efforts from resource-rich to resource-poor countries, autonomously functioning pediatric plastic surgery initiatives may also emerge in resource-poor countries. The GSR Institute of Craniofacial Surgery in Andhra Pradesh, India,⁵⁷ demonstrates that local physicians may develop a subspecialty plastic surgery center with a multidisciplinary team. The founding surgeon was the initial surgeon and administrator on the team and set up the cleft center in an existing hospital that was strategically situated between multiple states with large cleft populations. Additional team members in pediatrics, otolaryngology, speech therapy, neurosurgery, orthodontics, and dentistry were recruited in addition to a core group of nurses. Ultimately, the craniofacial institute became an independent center, relying on support from foreign and domestic funding sources and providing transportation, surgery, and nutritional counseling for many patients.

WORLD HEALTH ORGANIZATION DISEASE CONTROL PRIORITIES

The majority of global surgery outreach has been led by small groups, academic centers, and non-governmental organizations, and only recently has WHO formally recognized the important role of surgery in public health at an international level.⁵⁸ Although grassroots initiatives and efforts made by motivated volunteers are invaluable, global leadership in defining disease control priorities is important for raising awareness of the unmet surgical needs of resource-poor countries and garnering financial, legislative, academic, and public health resources that can help facilitate change.⁵⁹

WHO is well suited to bring surgical care to the global agenda. As an agency of the United Nations assigned the task of coordinating and promoting international public health, WHO has the ability to establish policies and coordinate resources from its 194 member states and outside funding mechanisms that may bring about widespread change.⁶⁰ WHO, in collaboration with other major entities, including the World Bank, Fogarty International Center, and Gates Foundation, published the second edition of *Disease Control Priorities in Developing Countries* in 2006,² which for the first time had a chapter on surgery. Congenital anomalies, including cleft lip and palate, and traumatic injuries, such as fractures and burns, were listed among a group of common surgical conditions responsible for a significant burden of disease.

The burden of surgical disease, of course, goes well beyond pediatric plastic surgery to involve a wide spectrum of acute surgical problems that low- and middle-income countries often lack the resources to address. WHO has tried to intervene through its Emergency and Essential Surgical Care Project.⁶¹ The goal is to engage general physicians, nurses, technicians, and paramedics in basic surgical techniques that can be performed at primary care hospitals to help meet the demand for surgical care. Future policy planning will be critical in defining how training and services are delivered to underserved populations, and pediatric plastic surgeons have an important role in formulating global surgery strategies.

Efforts are already underway to try to determine which surgical needs can most practically be met at a global level when taking into account surgical complexity and necessary resources. Considerations include equipment requirements, procedure complexity, and the need for supportive services, such as a blood bank, pathology laboratory, and intensive care unit. One strategy has been to prioritize surgical conditions by the extent of the public health burden, the availability of a surgical procedure that is highly or moderately successful, and whether the procedure is cost-effective and feasible to promote at a global level.⁶² In this hypothetical scheme, cleft palate repair was assigned a priority of 2, which falls into the intermediate category. Reasoning for categorization of various procedures was not provided, but this may have been related to the requirement for scarce resources, including orthodontics and speech therapy.

Surgery is receiving more attention on the public policy and planning stage than it has ever received before.⁶³ The reality of global surgery is that health for all people must be maximized with limited resources. This is an exciting opportunity for pediatric plastic surgeons to contribute to multidisciplinary discussions about disease pathology with which they are familiar, ranging from trauma to burns and congenital craniofacial and extremity anomalies. By sharing insight into disease management strategies and volunteering plastic surgery knowledge and experience, we can help improve global health.

CONCLUSION

Congenital anomalies, extremity trauma, and burns are among the most common surgical challenges facing resource-poor countries, and plastic surgeons have the knowledge and technical skills to manage many of these problems. Successful global surgery initiatives require that volunteer surgeons not only be well prepared to deliver care, but also find ways to collaborate with local systems. The most successful global surgical programs have enabled a transition of project management and service delivery to local health care providers. Cost analyses suggest that it makes financial sense to pursue global surgery efforts to both improve peoples' quality of life and enhance their productivity and contributions to society. As the challenges of global surgery become prominent on the global health agenda, ongoing research into health care delivery systems, local team building, personnel training, and resource allocation will be fundamental to expanding the availability of global surgical services.

KEY POINTS

- Surgical outreach has historically been omitted from global health agendas, possibly because the focus has been on communicable diseases.
- Pediatric plastic surgeons have an integral role to play in the variety of disease processes regularly encountered in practice.
- Global health programs have often been distinguished as representing a vertical or horizontal model of health care delivery.
- The potential academic and training benefits of global health trips are to health care providers from both volunteer and recipient countries.
- Evidence exists that pediatric plastic surgery initiatives can take root in underserved areas and become autonomously functioning entities.
- The majority of global surgery outreach has been led by small groups, academic centers, and nongovernmental organizations, and only recently has WHO formally recognized the important role of surgery in public health at an international level.

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3

How to Approach, Evaluate, and Treat the Pediatric Patient

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valuation and treatment of children as plastic surgery patients is a rewarding but often exasperating experience, one that can require an exceptional degree of patience. As in pharmacology dogma, children and infants should not be considered merely small adults. Developing a supportive physician-patient relationship with children is not simply a minimalist, scaled-down approach. Although treatment of adults has its own nuances, adult patient care is characterized by mature behavioral styles and advanced knowledge and reasoning capabilities. In addition, most adult patients are capable of self-consent. Children, however, require a more elementary approach to engage and educate them concerning their treatment. Their reasoning capabilities are usually immature, and they lack knowledge of the most general aspects of health care and well-being.

In this chapter we offer one approach to the evaluation and treatment of children. Although a successful physician–pediatric patient relationship is as individual as the provider and the patient, in this chapter we introduce some insights that may be helpful in treating pediatric patients.

THE FAMILY UNIT

The Child's Support Structure

First, the *family unit* must be defined. Treatment of pediatric patients and their families is unique. The family should be treated as a composite unit, which includes the child patient, parents and/or guardians, and sometimes siblings. The physician must also form an individual relationship of trust with the child. Parents and guardians have varying degrees of sophistication and success in coaching children through the medical process. Demonstration of parenting styles with regard

to behavior modification, discipline, and reactions to stressful situations is magnified in the reaction of the child. For instance, most of us have seen supportive and calm parents who were able to assist and guide their child through the discomfort of a needlestick and closure of a laceration, and then encountered other parents who were not able to provide similar support for a minor examination, resulting in extreme distress for all involved. The cooperativeness of the family unit can be extremely mature and is amazingly complex; it changes over time during subsequent visits, family stressors, and with the age of the patient.

Interaction styles and ultimate decisions on recommended care are somewhat dependent on a child's home situation. The primary caregiver during the day may be the mother, father, guardian, or another child care provider, all of whom are involved in the success or failure of a proposed treatment. Unlike adults, children must always rely on someone else (a guardian) for administration of medications, wound checks, care of incisions, dressing changes, positioning, and other restrictions. Responsible (and sometimes irresponsible) family members greatly affect the treatment outcome. Therefore it is imperative that physicians understand the unique family dynamic, the parents' educational and functional level, potential conflicting interests (such as those of siblings), and the living situation to provide the best care. Appreciation of a family resource pool, adjunctive services, and time constraints, particularly in single-caregiver families and those with other children with special needs, must be considered.

A second potentially conflicting factor in pediatric patient care is the socioeconomic status of the family unit. Children make up the largest indigent population in the United States. The largest demographic in Medicaid enrollees are children, and Medicaid served 30 million children in 2014.¹ No other large group of individuals has such a vast potential for disparate care. Numerous additional children qualify for Medicaid but have not been enrolled and are not insured; the U.S. Census Bureau, before implementation of the Affordable Care Act, estimated that this group comprises another 8.2 million children.¹ Children living in poverty and lower socioeconomic conditions have numerous direct issues of concern. In addition to being at higher risk for chronic conditions such as asthma, they are less likely to have appropriate immunizations and well-baby checkups, and their visits to the emergency room are more frequent.²

Potential problems for plastic surgery patients include an inability to purchase dressing supplies (often not covered by medical assistance programs), working-poor parents who cannot afford to take time off from work for the medical care of a sick or postoperative child, and the limited number of providers who will treat uninsured patients or patients receiving medical assistance.

An additional problem of the financially limited population is malnutrition. Improvements in social support and nutritional guidelines in children include the Women, Infants, and Children (WIC) program, Birth to Three programs, the California Children's Act, and individual state-mandated school cafeteria improvements, particularly in providing breakfast and lunch. Although most hungry adults are able to ask for or prepare food for themselves, in an immature child, hunger and malnutrition may more gradually become evident, as demonstrated by wound-healing problems, delayed growth, behavioral issues, and developmental delays. Counterintuitively, lower socioeconomic class is also associated with major increases in the incidence of childhood obesity because of the wide array of inexpensive, high-calorie, processed foods available. Obesity in childhood is linked to early-onset hypertension and type II diabetes mellitus, which affects surgical risk and outcomes.

Cultural Differences

Surgical outcomes are also influenced by the parents' or family unit's religious and ethnic beliefs. Respect for culturally diverse pediatric patients is essential to prevent alienation of these children

and their families and to best serve their needs. Although many religious and culturally diverse populations exist, we will briefly focus on the following four groups: Jehovah's Witness, Hmong, Amish, and American Indian.

Jehovah's Witness Families

The children of Jehovah's Witness families require special consideration. In this unique population blood transfusions are not promoted, and in some cases they are prohibited because of specific beliefs in biblical scripture. Although legal considerations must be met, the prevailing interest in the welfare of the child must be tempered with consideration for parental desires. Conflict with the religious wishes of Jehovah's Witness families may guide parents to decline treatment or seek acceptable (and controversial) alternatives, such as preoperative treatment with erythropoietin for an upcoming operation with a potential for high blood loss.³ Furthermore, some groups do make exceptions for children, allowing Cell Saver autologous transfusions, although this exception is infrequent.⁴ In cases in which a child's life is in danger as a result of massive blood loss, the courts have consistently supported physicians who transfuse without parental consent, siding with the overall outcome of life preservation and protection of the minor child. It is best to have frank pretreatment discussions regarding the family's wishes for transfusion, to consider alternatives, and to enlist legal intervention when necessary.

Hmong Families

Other ethnic groups such as Hmong immigrants have strong beliefs about reliance on traditional Eastern healing and herbal remedies and a general distrust of Western medical practices and surgery. Children may present late with complications or after failure of other methods, as evidenced by delayed presentation of appendicitis with very high rates of perforation in this population.⁵ Traditionally, illnesses in the Hmong culture are divided into spiritual and nonspiritual causes. Nonspiritual causes are thought to include illness from the environment, food, or drink. Spiritual causes include "loss of souls." Souls are believed to govern the body and maintain health. Soul loss can occur as a result of illness or any invasive procedure such as surgery, phlebotomy, or dental fillings. Touching a child's head may also release these good spirits; therefore a friendly pat on the head in this population is discouraged. In the Hmong belief system, shamans have the power to retrieve lost souls and restore health through ancient practices such as pinching, cupping, and coining to draw out fever, relieve headache, and alleviate vomiting. Western medicine providers may inaccurately interpret marks on the skin left by such maneuvers as the result of physical abuse. Loss of souls is thought to occur with surgery by allowing good spirits to leave the body and bad spirits to enter.⁶

The Hmong generally believe in reincarnation. Basically, if a person dies without all anatomic parts intact, in the next life, the person will be born with missing tissues (that is, cleft lip and palate, scars, and other deformities).⁷ Sensitivity and knowledge of these particular practices are essential when providing informed health care to Hmong patients.⁸

Amish Families

The Amish (who call themselves "plain folk" rather than "Amish") have a unique set of cultural beliefs that influences attitudes and utilization of modern medical practices. The Amish are conservative Protestants who strongly believe in pacifism, separation of church and state, peaceful living, and hard manual labor. Advanced education is not routinely encouraged. Hard manual labor jobs and reluctance to use modern conveniences such as electricity, telephones, and gas-powered transportation are typical. Reliance on non-Amish neighbors and friends is often necessary for transportation to health-related appointments. The Amish use home remedies such as mustard pastes and natural homeopathic medicines. Members of this culture routinely refuse social

financial assistance, health care plans, and health maintenance organizations. Decisions regarding medical and surgical treatment in individuals are made by the community leadership, governed by elders and lay leaders within the Amish community. This group routinely pays for care from community funds and will seek professional medical treatment only if the community agrees.^{9,10}

American Indian Families

More than 558 separate American Indian nations and tribes are recognized within the United States. Health care delivery to these unique populations should respect the cultural, historic, genetic, and spiritual diversity within these groups. In addition to increased awareness and sensitivity, health care delivery must also focus on the general health concerns of this population, including the growing epidemic of obesity. For example, in a 2003 survey of 263 Hopi children 6 to 12 years of age, 23% were classified as overweight and 24% as obese.¹¹ Infant mortality rates are also significantly higher in American Indian populations, compared with those of age-matched control groups. Lower socioeconomic status and higher rates of parental alcoholism, fetal alcohol syndrome, diabetes, and unemployment are believed to be contributing factors.^{12,13} The U.S. Census Bureau states that among American Indians and Alaskan Natives, 27% of children are living below the poverty level.¹⁴

Health care delivery can be complicated by subtle social differences. One American Indian physician from South Dakota discussed these cultural differences, specifically, time management and physician-patient eye contact.¹⁵ For example, a common provider misperception is that families and patients are disinterested because of a lack of direct eye contact and tardy arrivals or missed appointments. However, in some Indian cultures, direct eye contact is perceived as a sign of disrespect, and clock-watching is discouraged. Lack of reliable transportation and phone services in some groups compounds this problem.

PARENTAL PERMISSION, ASSENT, AND INFORMED CONSENT

Children do not have the ability to provide informed consent. Informed consent can only be provided by a person with the intellectual capacity and the legal right to do so. A child can, however, offer his or her assent to a procedure or treatment, thereby giving permission. Legally, even though a child gives permission, the responsible adult must provide permission.¹⁶ Obtaining a child's assent is encouraged, because it helps to involve the patient in the process of medical decision-making. A child's ability to provide assent varies, depending on age. The process of obtaining assent also varies, depending on the child's age and ability to comprehend the medical decision-making process. Exceptions to this rule include cases in which a minor is legally emancipated, or when sexually transmitted diseases, pregnancy, or substance abuse is at issue. When parental permission and assent are withheld, and the health of the child is at risk, arbitration or even adjudication may be needed.¹⁶ In acute life-and-death circumstances (such as those requiring blood transfusions in minors of the Jehovah's Witness faith), the courts have supported physician action to prolong the life of the child.

REACTIONS TO TRAUMA, CONGENITAL ANOMALY, OR DISFIGUREMENT

Patient and Parent/Caregiver Anxiety

Many patients in a pediatric plastic surgery practice have significant outward disfigurement and disability. Parents are frequently asked inappropriate and nosy questions about their child's condition by uninformed but well-meaning and curious family members, friends, and strangers. Many parents are quite anxious, distraught, and guilt ridden, somehow believing they are responsible for their child's condition. In addition, in the newborn period, parental depression is common,

with the realization that the infant is not “perfect.” Providers must spend significant time offering compassionate counseling to parents, which often includes meeting with child psychologists or social workers to help with strategies for managing questions and unwanted social attention.^{17,18} Reduction in the parents’ anxiety and guilt will ultimately be reflected in the child’s acceptance of the condition and surgical corrective procedures.

Another interesting phenomenon is the parent’s acceptance of the deformity.¹⁹ In our experience, parents have frequently expressed sadness around the time of a cleft lip repair, stating that they miss the cleft and the baby’s initial appearance. We have also witnessed the extreme of this adjustment, when a mother who did not believe the infant with a newly repaired cleft lip was her son. She was distraught until she heard the familiar cry of the infant and realized that the child was indeed hers.

Some parents are better able than others to handle discussions of medical procedures. For parents who are extremely anxious and queasy, extra time for discussion, a review of realistic postoperative photographs, and meetings with parents who have lived through the experience will provide some of the best solutions.

Preconceived Beliefs About Plastic Surgery

Beliefs about what plastic surgery can accomplish are greatly exaggerated in the popular media, and many parents’ expectations are unrealistic. For example, the most common discussion in the pediatric plastic surgeon’s office is about scars. Parents (and child patients) often have unrealistic concepts of scars, believing that somehow a scar can be completely erased. The reality of scar permanence and the time it takes for complete scar maturation is distressing to a misinformed and immature patient. Considerable time and local scar manipulation can make most scars less noticeable (that is, flat, similar in color to the surrounding skin, and nonirritated or nonpainful), but a scar resulting from reconstruction or traumatic injury will always be present. This fact must be repeatedly reinforced. For example, in our practice, removal of extremely disfiguring darkly pigmented nevi on a child’s face or body has often resulted in postoperative distress with the realization, “There’s a scar!” It is very helpful to revisit the preoperative appearance in photographs to help parents and patients recognize the improvements that have been made. The goals of any surgery should be realistically and repeatedly discussed with the child and parents to prevent an unrealistic expectation of undeliverable results.

Surgery for correction of a cosmetically significant deformity is frequently necessary and is not trivial. Payers, health maintenance organizations, and medical assistance administrations are often confused about this reality. Revision of a significantly asymmetrical cleft lip repair will not improve the function of the lip (usually) or provide a significant functional life-survival benefit, yet that does not mean a revision is an unnecessary procedure. Correction of an obvious deformity is essential for appropriate facial balance and development of appropriate self-perception and psychosocial well-being. This adjustment and the value of surgical correction have been extensively studied in the cleft palate population.²⁰⁻²³ Removal of a disfiguring pigmented nevus on the face of a child may be necessary for optimal socialization, even if removal does not improve the survival of the child. Numerous outcome studies have investigated facial disfigurement and the ability to achieve optimal socioeconomic status, educational level, and well-being.²⁰⁻²³ Many children and adults with disfiguring conditions have overcome this stigma. These patients are exceptional in their abilities, energies, and social support system.

The nature of many plastic surgery procedures for children results in denial of coverage by third-party payers and health maintenance organizations. Insurance companies and governmental medical assistance programs are not currently federally mandated to cover reconstructive procedures for children with deformities resulting from congenital abnormalities, burns, and trauma. Pediatric plastic surgeons must frequently challenge insurance companies’ denials for coverage

of care, placing personal phone calls to medical directors, informing payers about pediatric plastic surgery conditions, and continuously pressuring legislators to pass laws requiring children's deformities to be covered by insurers.²⁴ Pediatric plastic surgeons frequently must educate the public and advocate for their patients who cannot advocate for themselves, both on an individual and a group level.

COMMUNICATION OF NEEDS AND DESIRED OUTCOME

Developmentally appropriate-level discussions regarding specific reconstructive goals are necessary, particularly with older children and teenage patients. A pediatric plastic surgeon may have a particular goal in mind (for example, reducing the size of the nasal tip and bony deviation in a cleft nasal deformity), whereas a teenager may be more focused on the scar. Although it is an unusually sophisticated child or teen who can self-analyze an unacceptable nasal contour, providing a hand mirror during the discussion and acknowledging the child's and the parents' concerns will ensure everyone is in agreement in defining goals for surgery. Preoperative and postoperative photographs of children with similar features are helpful to discuss goals, and particularly to emphasize what can and cannot be accomplished with plastic surgery. Usual postoperative care and expectations should be reviewed in detail. For example, in the case of rhinoplasty, to an adult the idea of postoperative swelling, the presence and sensation of intranasal packing, an expectation of minor bleeding, and the need for a dorsal splint may seem simple concepts, but for a child these events must be explained in detail. In addition, children usually require frequent reassurances and reminders that what they are experiencing is normal.

Surgeons should not take any criticism too personally when children view their early postoperative results. For example, a child who underwent subtotal nasal reconstruction for Binder syndrome (nasomaxillary hypoplasia) was teary and despondent during the unveiling of her newly reconstructed nose. She wailed, "It's so big, it's so big," because she was familiar with only very little nasal contour, and the result was shocking to her. Although similar outbursts can occur with adult patients, usually dissatisfaction or the early reaction to the result is more controlled, subdued, and reasonable. Additional time and consolation is needed for pediatric patients who are shocked or disappointed with the early results. The nasal contour result in the Binder syndrome patient was beautiful, and by her second postoperative visit she had become accustomed to the look and was ecstatic. By her own admission, it was such a drastic change that she was distressed by it.

PSYCHOSOCIAL IMPACT ON SELF-ESTEEM AND WELL-BEING

The current cultural emphasis on outward beauty and aesthetic norms has had a significant impact on children and their perceptions of beauty and acceptance. It is interesting, for example, that as early as 3 years of age, children with cleft lip identify the cleft in self-portraits. The significance of a scar on the upper lip is the same as if daddy wears glasses or mommy has long hair. However, by the time these children are 6 years of age, the cleft lip scar has resulted in psychosocial negative attention and affected their sense of well-being. Numerous potentially abnormal appearances, such as protuberant ears, asymmetrical lips, visible pigmented nevi, and contour irregularities of the craniofacial skeleton are treated within this critical time period, and frequently surgery is performed between kindergarten and first grade to minimize the psychosocial impact of the deformity.²⁵ Children require special attention if they become withdrawn, get into fights, or are wary of school, because they may be receiving negative attention at school or in other social interactions as a result of their deformity. Child psychologists, social workers, clergy, parents, teachers, and other caregivers can help children through the difficulties of growing up with scars and deformities. A close support structure helps children to develop strategies for coping

with negative attention or teasing.²⁶ As part of the history-taking process, the provider should ask whether a child demonstrates aversion to or anxious behavior in school and other social situations. Children who are more mature can be asked directly if they receive negative attention, and their coping strategies can be discussed to uncover problems in this area. A child who experiences significant teasing and negative attention will be forthcoming if the physician has first established a strong and well-developed trust.

ESTABLISHING A PHYSICIAN-PATIENT RELATIONSHIP

The Physician's Demeanor

A pleasant, happy, and somewhat gregarious demeanor rapidly wins over even the most timid and withdrawn patient. Nowhere in medicine is it more important to engage patients with direct eye contact and to speak directly to them (not to the parent or guardian). It is often advantageous to know the latest cartoon characters, movies, and toys. Children will open up if they find the provider interesting and interested in them. Silliness is unnecessary in most interactions but may be necessary with a particularly withdrawn child. For example, one of our colleagues, when meeting with a particularly troublesome child, will make a bet with the child that he has a fish in his pocket. The child usually states, "No you don't." Bantering back and forth leads to our colleague's displaying his fish-shaped writing pen from his pocket, and he continues to make his notations with the fish pen. He even lets the child hold it in the preoperative photographs. This method sets the patient at ease, engages the child with the surgeon, and provides a good laugh during the examination.

In interactions with children the surgeon should never reveal a "bad day" mood, an "I'm running an hour late," hurried behavior, or an "intellectual curiosity" look of consternation or bewilderment when thinking, "Now, how am I going to fix this?" Children directly reflect and communicate their fears, apprehension, and stranger anxiety.

Effective Verbal Communication

Continuing to talk during the examination is crucial. Children become less anxious when they hear the provider's voice, even if the topic is soccer, cartoon characters, or a favorite candy flavor. Surgeons should explain what they are going to look at, why they need to look at it, that it will not hurt, and that they are going to use a flashlight. Waiting for an invitation to examine is counterproductive, but explanations should proceed at the time of the examination. A child who seems timid or scared can be allowed to examine the surgeon first by shining the light in the surgeon's or parent's mouth to understand the examination.

Saying "thank you" to children works wonders (for example, "Thank you for letting me look in your mouth. That was very kind of you. Your tonsils look great today!"). This behavior reinforces a child's involvement and self-consent for care. Appreciation of the child's participation will establish a positive overall tone for the day's visit, benefitting the physician and the child, and will help to ensure a mutually cooperative mood at the next visit.

Child Empowerment in Decisions Regarding Care

With cantankerous children, we often explain what we need to see and then let them choose the order of the examination. Sometimes we examine patients' siblings, parents, and medical students in the room before the patients allow us to examine them. We ask if they would like a special Band-Aid, allow them to choose the color, and ask if they would like cherry- or grape-flavored medicine. By empowering them with even the most trivial of decisions about their care, children think they are active participants in their care and are satisfied to be involved in the team.

White-Coat Anxiety

Children sometimes cringe or cry immediately when they see a physician in a white coat. This phenomenon is not unique to children; we have all heard about hypertension related to “white-coat syndrome.” Children may have anxiety or distress from prior painful interactions (for example, immunizations) and unconsciously relate the white coat to painful stimuli, although this relationship is controversial.²⁷ It is rarely necessary for the physician to remove his or her white coat in the examining room for the patient’s comfort. We prefer not to do this, because we believe the advantage of a white coat, which delineates our role as physicians, is important. However, at our University Hospital, the current pediatric teaching is to go “coatless” to not cause anxiety or stress in the children. We think the white coat symbolizes that the person wearing it is to be trusted, similar to the effect of a police officer’s or firefighter’s uniform. The coat helps a child understand that an examination will occur. It eliminates confusion about being touched by strangers (that is, it is acceptable for a person in a white coat to touch and examine the child, even in a sensitive area, but a stranger without a white coat should be discouraged from touching).

EXAMINATION

Eliciting Cooperation

Children can be notoriously uncooperative with a painful or uncomfortable examination, and often solicitation to proceed is required. Older children can usually understand a simple explanation of why it is important to examine the area. However, reasoning does not work well with a very young or immature child, who may spend endless time asking, “But why?” Offering a reward often succeeds in encouraging cooperation in an immature child. Most pediatric plastic surgeons keep well-supplied award boxes filled with small rewards such as stickers or pencils. Many parents will also help with rewards, such as a promise of lunch at a restaurant, the purchase of ice cream treats, or toys. However, rewarding a child for cooperation does not work well for those who constantly have this type of reward tactic used at home. If parents frequently reward children for every activity, the value of a physician’s reward at the office is significantly diminished. These parents are easy to identify: they usually arrive at the clinic with multiple toys, security objects, and food items. In these instances we reward patients with extra attention and compliments, such as telling them how much we like their shirt, admire their bravery, or like their hairstyle. When praising a child for cooperation, words such as “good” are not used, because they are too general. We tell them we admire how brave they were, how grown-up they behaved, or that we wish all of our patients were like them. Sometimes the best reward is a sincere compliment; even very young children can detect false praise.

Sequence of Examination

The sequence of the examination should be logical, proceeding from least intrusive to most intrusive procedures. For example, if a child has a laceration to the arm, the physician should not start poking the arm right away. The arms are examined while they are outstretched together. The physician then listens to the child’s lungs and heart, and once the child is comfortable with the examination, focuses on the affected area. Children develop a trust in the examiner’s touch during the examination of the nonpainful area. It is sometimes helpful to allow parents or the children themselves to remove bandages. The use of blunt, nonthreatening-appearing bandage scissors and liquid adhesive remover is essential, because it facilitates painless removal of sticky bandages and tape.

Head, Neck, and Oropharynx

In small children and infants, we prefer to perform examinations of the head with the child lying in our lap and looking at mom or dad. Not only does this provide a better perspective for evaluation of head shape anomalies, but it also allows the infant or child to face the parent. Most infants are unable to lift their heads easily; therefore accurate head circumference measurement and clinical examination of shape are enhanced. The oropharynx can be easily examined in this position. The physician can make a game of going upside down, and even if the child becomes unhappy and cries, the view of the intraoral structures is optimal.

Tongue depressors are unnecessary for examining most children. It is always evident when a child has previously gagged on a tongue depressor. Even the use of flavored sticks (grape and cherry) cannot overcome a child's memory of discomfort caused by gagging. If a tongue depressor needs to be used, the child is never allowed to keep it because of the risk of aspiration or injury. Instead, the physician can drop it on the floor and claim "no 5-second rule here," or quickly discard it and refocus the child's attention on another object. Tongue depressors can be very dangerous to young children and should always be carefully discarded.

Children are exquisitely ticklish, so rapid examination of the neck for lymphadenopathy or torticollis should be avoided. Slow and gentle pressure using the fingertips will reduce the tickle discomfort.

Body and Trunk

Most children have little difficulty with body and trunk examinations. Although children in general have less phobic attitudes toward nudity, areas already examined should be shielded or covered. Children deserve no less respect for personal privacy than adults do. In preteen and teenage patients, extra consideration for privacy is warranted. These patients are extremely self-conscious about their bodies.

Hand

Examination of the hand will be thoroughly covered in other chapters, but we will mention here that observation is essential. A child will reach for toys or hold the hand in a cascade posture. Sometimes a game of "Simon Says" can help with diagnosis of hand function. In addition, tendon integrity can be assessed with nonpainful pressure on the upper forearm (far away from the injury in a lacerated finger) and observation of the finger movement and posture. We have found this to be very effective with anxious and uncooperative children in the emergency room.

Other Examination Pearls

Low, Calm Voice

A low, calm voice is essential in an examination and interview. A calm, cool, and collected demeanor will be reflected in the behavior of the patient and parent.

Management of Tantrums

Occasionally, a patient will have an all-out, down on the floor, kicking and screaming tantrum. The physician should calmly restate what will be done and let the parents attend to the child. This method rarely fails. Sometimes everyone in the room needs a time-out—to collect themselves and restart the examination.

CHILD-CENTERED ENVIRONMENT

Visual Stimuli

One of the most elementary comforts for children is being in a child-friendly environment. The offices of most pediatric plastic surgeons are significantly childproof. The office should have obstructions to child's play with computers or instruments, locking drawer systems to eliminate pinched fingers, and light switches and door handles out of reach. For the toddler room, padded corners of tables and desks will reduce the risk of head injury in a walking child.

Children become restless sitting in a small examining room and should be escorted into the room only immediately before the physician enters. In our practice, children and families wait in a large waiting room complete with movies, computers, and toys where they can play and interact with other children. Time waiting in an examining room is minimized. In addition to being childproof, examining rooms with colorful and fun wall hangings, activity centers, mobiles, and toys improve the child's enjoyment of the office visit. Toys in the room should be thoroughly cleaned or replaced between patients and should pose no risk of aspiration (that is, have no small or removable parts).

Children respond favorably to child-sized furniture. They will feel more comfortable if they are able to sit in a child-sized chair or stool rather than awkwardly climbing onto an adult chair with their feet hanging over. Child-sized furniture makes patients feel important and reinforces the perception that the clinic is for them.

Employment of "childcentric" staff is an absolute necessity. Staff members who become annoyed with childish behavior should be reassigned to adult clinics. All staff, including nurses, clerks, physician assistants, and medical assistants, should have a focused interest in the treatment of children.

DAY OF SURGERY

Anxiety Reduction

There is no ideal way to totally eliminate tension and anxiety on the day of surgery. To help, however, a visit by the pediatric plastic surgeon before the patient enters the operating room is essential. Familiar faces, such as nurses from the clinic, will also help to place the entire family group at ease. Before entering the operating suite, most toddlers and children will benefit from some type of mild oral anxiolytic agent.^{28,29} In our institution, the "goofy juice" (usually midazolam and acetaminophen) is well received by children (who are thirsty) and by parents, who are comforted when they see that their child is relaxed before the procedure.³⁰ The preoperative room has child-centered decor, cartoons on the television, and child-appropriate movies.³¹ We believe the presence of the parents on induction of anesthesia is essential to a pleasant operative experience, and only rarely do parents decline.³⁰ Child-life specialists accompany the parent and child into the operating room; they explain the events and are experts in counseling and reducing anxiety in parents and children. They then escort the parents to the waiting room after the child is anesthetized and provide frequent updates from the operating room and supportive care to the parents during the operation.

Intravenous lines are placed, and painful phlebotomy is performed only after the child is asleep and the parents have left the room. Mask induction of anesthesia, which is used in infants, toddlers, and young children, is flavored with cherry, grape, or another flavor. In older children, copious amounts of topical anesthetic cream are used (for example, EMLA) before an intravenous stick. At our institution, intravenous anesthesia is used for older children and teens. There is no age limit for parental presence in the operating room, although most teens self-select to exclude parents from the operating room. Calm and enjoyable family experiences are influenced by

our pediatric anesthesiologists, who often tell bedtime stories to the children as they undergo induction of anesthesia. This sensitive care is appreciated not only by the child but also by the parents, who understand that we are a team dedicated to the care of children.³⁰ Feedback from parents has been positive and is also evidenced in children who must undergo multiple operations and are not anxious at return visits.

POSTOPERATIVE COURSE

Pain and Anxiety Management

When the child arrives in the recovery room and is still sleepy, the parents are escorted in by the child-life specialist. At no time does the child sense that the parents are not present. The child-life specialist remains with the family to explain events and provide support. In the recovery room, pediatric-specialist nursing care is essential. Many children and all infants and toddlers cannot effectively verbally communicate discomfort. Nursing specialists in pediatrics are specially trained to watch for subtle signs of infant and child discomfort and are experts in giving appropriate pain medication, including narcotics. Pediatric nurses facilitate immediate interaction with the parents, including activities such as play or allowing parents to hold the child in a rocking chair. Children who need to be admitted to the hospital after the procedure are housed in a room with childcentric decor with parental rooming-in capability. Support in the preoperative area by trained child-life specialists and play therapy in the postoperative period have been shown to reduce child and parental anxiety and speed recovery.^{32,33}

KEY POINTS

- Children are not just small adults.
- Children account for a disproportionate segment of the indigent population.
- Low socioeconomic class has the potential for a negative impact on the following:
 - Nutritional status
 - Access to general well-child care
 - Access to other health care resources
- Although ethnic and cultural beliefs influence the care of children, health care professionals must understand individual family concerns and background, and decisions must be made that are in the best interests of the child. This may require enlisting an independent child advocate or the court system.
- A child-centered environment, including kid-friendly health care professionals, reduces anxiety in a pediatric patient undergoing surgical treatment.
- Insurance companies and governmental medical assistance programs are currently not federally mandated to cover reconstructive procedures for children with deformities resulting from congenital abnormalities, burns, and trauma.

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4

Ethical Issues in Pediatric Plastic Surgery

John A. Persing • Elizabeth G. Zellner



Although technical skill and proficiency are key qualities of a successful surgeon, the choice of when to operate, and perhaps more important, when *not* to operate, is one of the most critical decisions that a surgeon ultimately makes. Virtually all plastic surgeons, along with the rest of our medical colleagues, took some iteration of the Hippocratic Oath on graduation from medical school. Although passages regarding not “cutting for stone” have largely become historical curiosities, most of us remember the promise to “first do no harm.” Ironically, this phrase was not part of the original Hippocratic Oath, but rather is attributed to Thomas Inman, a nineteenth century surgeon.¹

This key medical premise is inherently challenged by surgical procedures, including those in plastic surgery. As with any surgery, there is a period of potential perioperative “harm” done to patients as a result of surgical trauma. When this harm is done to remove a cancerous tumor or restore a working limb, the temporary pain and discomfort of the intervention are quickly discounted as necessary and unavoidable. However, when the desired change is not a lifesaving procedure but rather a functional or cosmetic improvement, the issue becomes less distinct. Although every plastic surgeon strives to be technically capable and well informed and to perform procedures safely, no surgical intervention is without some risks. Even benign procedures can, in special circumstances, be lethal.

In this chapter we provide a basic framework of the ethical issues inherent to medical practices and to identify some challenges unique to pediatric plastic surgery. As technology and societal expectations continuously evolve, only a sound adherence to ethical principles and sincere attempts to work toward patients’ best interests can truly guide a surgeon through unique and often ethically complex dilemmas.

The American Board of Medical Specialties identifies six core competencies for all practicing physicians to learn and master during training: patient care, medical knowledge, interpersonal and communication skills, professionalism, systems-based practice, and practice-based learning and improvement. Although much work is done on teaching the other core competencies, a recent search turned up only two articles within the plastic surgery literature that discussed training future surgeons in professionalism in a formal manner despite the fact that many plastic surgeons face complex ethical questions every day.² Chung et al³ searched the literature for any papers on ethical issues and found only 110 articles out of more than 100,000 searched that focused on ethical principles. Of these, only 40 represented original research. They also saw a heavy bias toward a focus on patient autonomy and beneficence versus the other main principles of nonmaleficence and justice.³

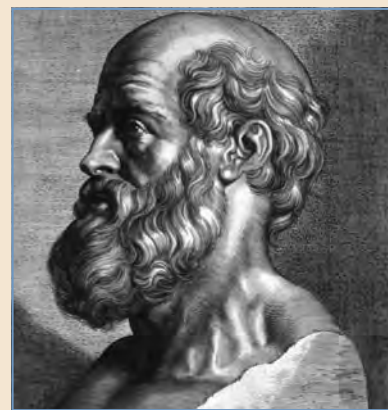
HISTORICAL PERSPECTIVE

Many of the most challenging issues that confront physicians involve how we view the pursuit of new knowledge and at what cost this knowledge may come. Modern medicine has evolved in some ways at the cost of some questionable previous practices: from medieval anatomists stealing cadavers for study, to the terrible atrocities of medical experiments that were conducted within the Nazi concentration camps, to the injustices of the syphilis studies at Tuskegee, Alabama. Through careful self-examination of our actions and duties as physicians, we must come to a point at which the patient-physician relationship remains a bond of trust.

The first mention of an unethical physician comes from ancient Greek mythology, when the renowned physician Asclepius accepted payment for bringing a person back from the dead despite religious and professional admonitions against performing this act. For his hubris, he was punished by being struck dead by a thunderbolt. Despite these regrettable actions, he was later resurrected as a god for political reasons within the Greek polytheistic world.

The Hippocratic Oath followed shortly thereafter in approximately 400 BC. The Oath was part of the Hippocratic Body, written by Hippocrates of Cos or one of his students⁴ (Fig. 4-1). This document emphasizes loyalty to the profession of medicine, doing what is in the best interest of the patient, avoiding harming patients, and an early mention of patient-physician confidenti-

Fig. 4-1 Hippocrates of Cos, creator of the Hippocratic Oath about 400 BC. (Engraving by Peter Paul Rubens, 1638. Courtesy of the National Library of Medicine.)



ality. By affirming a set of ethical principles, the medical guild of ancient Greece set themselves apart from a purely mercantile establishment. From this point onward, a distinction between business and medicine was firmly established.

By the fifth century AD, the first written code of ethics, the *Formula Comitatus Archiotorum*,⁵ was published in the Ostrogoth kingdom of Theodoric the Great in what is modern-day Italy. This doctrine focused primarily on the duty physicians had to deepen their knowledge base and share their knowledge and consultations with fellow physicians.⁵ Throughout the medieval period, Muslim, Jewish, and early Catholic scholars commented on the line between religiously acceptable treatment and what the medical profession actually did. Ishaq ibn Ali al-Ruhawi wrote the first book on medical ethics during this time: *Practical Ethics of the Physician*.⁶

In 1794 Thomas Percival,⁷ an English physician and author, published the first modern treatise on the topic: *Medical Ethics; Or, a Code of Institutes and Precepts, Adapted to the Professional Conduct of Physicians and Surgeons*, which he expanded on in 1803 (Fig. 4-2). In this work he asserted the moral authority and independent service of physicians while affirming the physician's duty to care for the sick.⁸

By 1847 the American Medical Association,⁸ coincident with its founding, adopted its first code of medical ethics, borrowing largely from Percival's work. This code has been updated numerous times throughout the years, most recently in 2001, and is available at the AMA website.⁸ Prominent theorist Joseph Fletcher has been cited as founding "situational ethics" in the 1960s and 1970s and was one of the fathers of modern bioethics. He professed that abstract moral laws should accede to whatever seemed most loving in any given situation. He addressed topics ranging from eugenics to abortion and physician-assisted euthanasia, stirring debates that remain active to this day.

In 1979 Beauchamp and Childress published *Principles of Biomedical Ethics*,⁹ which revolutionized the way that most physicians discuss medical ethics today. Their central premise centered around four principles that can be applied in a flexible and balanced manner to answer the many complex ethical questions that arise in modern medicine. These guiding principles are beneficence, nonmaleficence, autonomy, and justice³ (Table 4-1). Unlike other ethical theories, this set of principles was designed specifically to tackle questions in biomedical ethics.



Fig. 4-2 Thomas Percival, creator of the first modern code of medical ethics in 1794.

Table 4-1 Four Main Principles of Medical Ethics as Advocated by Beauchamp and Childress

Ethical Principle	Description	Example in Plastic Surgery
Autonomy	Acknowledge and respect a patient's right to self-choice and self-governance, free from interference of others and from limitations toward making informed decisions.	Cosmetic surgeons have come under scrutiny that some of their advertisements may be deceptive or intended for the solicitation of vulnerable customers; thus receiving full and informed consent from patients about surgical procedures is jeopardized.
Nonmaleficence	"First do no harm." Obligation not to inflict harm or adverse effects to patients as a result of absence of care.	Because all physicians take an oath to "first do no harm," plastic surgery often falls well outside the traditional goals of saving lives, treating diseases, and promoting health.
Beneficence	Obligation to prevent or remove harm while also promoting good by contributing to the welfare and acting in the best interest of the patient.	Many innovative surgical cases are unique, and surgeons are constantly amending procedures to try to find the best ways to achieve optimal results; however, some may have undesirable side effects. In a disease state, potential benefits versus the potential risks are weighed; however, it is often hard to justify whether an elective procedure on a healthy individual is worth the potential risk.
Distributive justice	Distributing benefits, risks, and costs fairly, equitably and appropriately; treating patients with similar cases in a similar manner.	Many Americans are without insurance or have insurance plans that cover very little. Certain reconstructive surgeries and almost all cosmetic procedures are rarely covered and therefore only financially feasible for the affluent.

NONMALEFICENCE

Nonmaleficence is the only one of the four principles mentioned within the classic Hippocratic Oath, and most physicians find it the most memorable. As physicians, we have dedicated our lives to improving human health, and most try to complete this precept without inflicting harm to patients. However, within the realm of surgery, this is inherently somewhat inconsonant, because all surgery begins with physical trauma to the patient. One "harms" in order to cure. One cuts in order to improve.

Surgeons must constantly balance the risks and benefits of surgical intervention. Any ethical physician will not willingly undertake a procedure that he or she does not believe will benefit the patient. However, it is rarely a decision that is so clearcut. When a patient's benefit will be measured in psychological comfort or a reduction in anxiety, it is more difficult for a clinician to quantify that improvement versus the calculated risk. However, even small cutaneous scars have been shown to have significant effects on patients' mental health years later.¹⁰

Occasionally, a plastic surgeon may be asked to perform a procedure about which he or she may not be fully comfortable. Patients, fueled by the intense media fascination with body shape, may sometimes ask for results outside the realm of responsible plastic surgery practice. In these cases, a prudent surgeon balances the expectations of the patient with his or her own moral com-

pass and technical skill set to outline a plan that will please the patient yet still do no harm. In most surgical consultations, all possible alternative interventions, such as psychological counseling and other supportive treatments, should also be reviewed as alternatives to surgical procedures.

Most pediatric plastic surgery is considered reconstructive surgery. Reconstructive surgery differs from cosmetic surgery in that reconstructive surgery seeks to restore an appearance or function that is considered abnormal to normal. By contrast, cosmetic surgery seeks to improve what is considered normal to beautiful, usually adhering to classical ideals of beauty. Some critics argue that cosmetic surgery to enhance a patient's appearance is at the periphery of the realm of medical practice. Seeing no improvement in patients' physical health and the potential for numerous complications, they believe that cosmetic surgery is "a most unusual medical practice,"¹¹ in the sense that it does not cure disease or reduce physical pain and caters to a patient in a sometimes businesslike marketplace. There are certainly some gray areas; however, most ethical plastic surgeons recognize that they frequently are able to help a patient who has either physical or psychological discomfort by addressing the patient's concerns through surgical treatment.

It is important to screen potential patients for possible psychological comorbidities. One can think of this in terms of identifying unrealistic preoccupation with the body part in question, sometimes indicating *body dysmorphic disorder*. In most situations, if the deformity is proportional to the patient's anxiety about it, there is less likelihood of a negative underlying psychological issue. It is also more likely that the plastic surgeon will be able to deliver a result that is pleasing to the patient when both parties understand the concept of incremental improvement: moderate deformity can be made near-normal in appearance, and normal appearance can be aesthetically enhanced to beautiful. However, it is difficult to take a significant deformity and improve it beyond the normal range to beautiful.

At the same time, it is often challenging to make significant changes without tradeoffs in terms of telltale scarring or differences that are socially notable. It is important that all patients, adult and pediatric alike, have realistic expectations and understand and accept the limitations of even today's plastic surgery before any procedure is undertaken.

In pediatric plastic surgery, there is slightly less patient pressure to push aesthetics beyond the limits of what is considered the societal norm. However, parents and some older patients themselves are conflicted about what is considered acceptable in reconstructive surgery when perfect is not achievable. They must be carefully counseled and supported when further surgery will not improve the ultimate result. A prudent surgeon recognizes this and will not be pressured into further futile operations that will not benefit the patient.

BENEFICENCE

Beneficence, the concept of acting in the patient's best interest to do "good" for the patient, naturally arises from the idea of nonmaleficence. This takes the idea of doing no harm to the next level. As a medical practitioner, Childress and Beauchamp argued, it is not enough to merely keep from harming the patient; one must make an effort to actually improve the quality of the patient's life.⁹

However, in plastic surgery, doing "good" can be difficult to quantify. Certainly, improving appearance and self-esteem seems to be a worthy goal. Patients can be in real psychological pain as a result of self-consciousness about their appearance. Studies have shown that physical appearance has an impact on the quality of life; taller individuals, with other variables controlled, make more money, implying greater occupational success.¹² More attractive photographs attached to identical resumes also increase interview rates significantly.¹³ Perhaps, some argue, we should spend our efforts on erasing these biases in society rather than shaping humans to fit them. However, this is easier said than done, because some biases about physical attractiveness are deeply ingrained with basic tenets of survival, such as selecting a mate based on pelvis shape and indicators of fertility.¹⁴

Regardless of these claims, it is increasingly important in plastic and reconstructive surgery to show the value of surgical interventions. Much recent work has been done on patient-reported outcomes data.^{15,16} As we find ways to quantify patient improvement in quality of life and other outcome measures, it becomes easier to validate the good that is being performed in plastic surgery. It also helps clarify ways to further our field so that we are focusing on the interventions that make the most positive difference to patients. In the future, the perspective gained from this research will be invaluable in counseling patients who are contemplating surgical interventions for themselves and their children.

AUTONOMY

Autonomy is one of the most challenging ethical concepts in medicine, and in many ways it is a thoroughly modern concept. In past eras, medicine was practiced with an air of paternalism in which the doctor was the expert who decided the medical course for the patient. However, since the latter half of the twentieth century, emphasis has increasingly been placed on patients being more engaged in determining their own medical care. In this scenario physicians serve more as counselors and guides than as the driving decision-makers of medical care.

In general, regarding competent, mentally sound patients, surgeons provide counsel but ultimately leave it up to the patients to decide which risks they are willing to undertake to obtain the desired benefits. We formalize this calculated risk-taking through the formal process of written informed consent. True informed consent is a difficult concept in and of itself. In theory, patients are presented with all of the possible benefits of a given intervention and are also told of all the possible risks of the procedure. Even without acknowledging the time restraints and educational barriers that exist in any busy clinic, patients often have preconceived notions of what surgery can provide, and their in-depth understanding of subtle medical nuances is limited. The fact that a skin incision always results in a cutaneous scar and therefore is always unavoidable comes as a shock to some patients and their families. The complicated mechanics, wounds, and aftercare of many pediatric procedures can be overwhelming and are often not fully understood by nonmedical parents.

When patients are minors, which by general legal definition is regarded as individuals under 18 years of age, additional ethical challenges are encountered. Children cannot legally consent for themselves, which leaves this burden to their parents. In the case of life-threatening conditions, the choice to pursue surgery may seem obvious to parents. However, when the desired result of the surgery is a change in the patient's physical appearance, the choice can be less clear. No surgical intervention is free of physical pain during recovery, and some larger reconstructive procedures may have a substantial amount of surgical morbidity. The burden of deciding to undergo surgery can often be significantly distressing to parents.

Although minors cannot legally consent, some advocates push for obtaining pediatric assent according to whatever capacity the pediatric patient meets. Medically speaking, children between 0 and 7 years of age are without such capacity and are unable to make any meaningful decisions about their own health. Children between 7 and 18 years of age have developing capacity, and obtaining at least verbal assent from these children can be a valuable endeavor in bonding with the patient and providing a solid framework for moving forward surgically. Minors have capacity when they are considered mature, such as high school seniors over 18 or "emancipated" minors, who have undergone legal separation from their family's legal authority. These young adults will generally also have legal capacity to sign consent forms despite possibly seeing practitioners with their parents. In addition, those over the legal age of majority, with physical or mental disability so severe they cannot care for themselves, remain legal minors under the care of a legal guardian for the course of their lives.¹⁷

In the face of decisions about their child's future—weighing the benefits of a more normal appearance versus the prospect of surgery on an infant or a toddler—parents often hesitate. Although parents have been shown to be motivated by a desire to do what is right for their children, the choice is not always clear regarding whether a particular intervention is going to really improve their child's life¹⁸ (Fig. 4-3). Some parents and former patients have expressed concern about the ultimate motivations for changing the child's appearance. Is the surgery truly to make the child feel better (many of whom are unaware of their differences at the time of surgery) or to make the parents feel better about the child? It is important to recognize the motivations that drive the desire to correct certain congenital abnormalities. A reasonable conception of physically “normal” involves thinking of a bell-shaped curve with respect to normal human differences. Individuals that fall within the majority of the bell curve generally will be perceived as normal, whereas those who are more than standard deviations away from the mean can be easily recognized as “other,” even by small children.¹⁹ The goal of reconstructive plastic surgery is to restore patients closer to the mean to avoid psychosocial stigmatization. Pursuit of further aesthetic improvement beyond the mean to produce psychosocial benefit has not been scientifically validated, and falls within the purview of cosmetic, not reconstructive, surgery.

The onus falls on the medical profession, often plastic surgeons, to provide appropriate guidance and counseling to parents of children with congenital anomalies. For some conditions, there are common positions on whether or not surgery should be undertaken during childhood. For example, a cleft lip should be corrected early to avoid functional and social deficits, whereas asymptomatic cutaneous hemangiomas should generally not be removed while they still have the potential to regress spontaneously. Many of these consensus positions have been solidified only through decades and even centuries of clinical experience and continue to evolve as new knowledge becomes available.

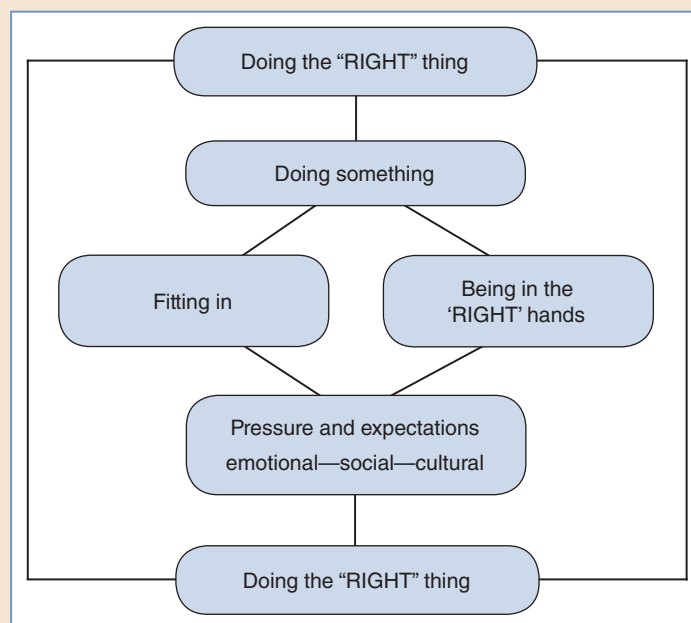


Fig. 4-3 Parental motivations can be complex when making medical decisions for their children.

In addition, organized medicine must continue to push the envelope in research on outcomes to quantify the value of proposed surgical interventions. Although a whole-vault cranioplasty in a child less than 1 year of age may sound too risky for a cosmetic improvement in the baby's head shape, the prospect of improved cognitive abilities and fewer learning disabilities makes this decision much more obvious to parents, who desire the best possible life for their child.²⁰

Although respecting minor patients' autonomy can be difficult under the best of circumstances, family dynamics can further complicate the picture. Legally, any parent with custody of the child can provide consent for surgery. However, at times, various parental interests may not be in agreement. In these situations, which occasionally may pit parent against parent, the clinician's personal sense of ethics and morality may be challenged. It may be helpful to involve the hospital's ethicist before proceeding with surgery in cases of conflicting parental wishes.

Certain special populations may offer further challenges to surgeons. People of the Jehovah's Witness faith usually refuse blood transfusions in the belief that this is in violation of biblical law. Before any surgery, this issue should be fully discussed with the parents, and plans should be made to minimize blood loss and the need for transfusion. In surgical cases with significant blood loss, whether predicted or not, the medical need to transfuse can become obvious. Even in cases in which parents will not consent to transfusion, courts have largely upheld the rights of physicians to transfuse a child (under 18 years of age or a legal minor) to save his or her life. This has been based on the protection of the minor child's life over the parents' religious beliefs. As mentioned in Chapter 1 on evaluating pediatric patients, other cultural differences, such as those found in Hmong, Amish, and American Indian societies, may additionally challenge the preconceived notions of physician ethics. It is important to be aware of culturally sanctioned differences in the perception of medical care and adapt accordingly to best serve these populations.

Finally, it is important to be aware of moments when the child's best interests and those of his or her parents are clearly at odds. Child abuse and neglect can exist, even where one is least likely to suspect such behavior. As physicians serving the pediatric population, plastic surgeons owe it to their patients to be vigilant with regard to possible abuse and to act in the patients' best interest. In any case of suspected maltreatment, physicians are legally required to report their suspicions to the appropriate child protective services.

JUSTICE

Justice is the final concept within the original medical ethics principles. Especially for children, who cannot dictate their family circumstances, it is important that safe and complete medical care be available to all, despite the ability to pay for care. Physicians have not been taught to think regularly of society's needs as a whole when faced with individual patient needs. However, as health care becomes increasingly regulated, this type of thinking and consideration will progressively become more important.

This is especially crucial in various congenital conditions. It has been argued that reconstruction of a hand should be performed while a child is still developing motor control to take advantage of the plasticity of a younger brain and maximize functional outcomes. Soft palate function should be restored before speech development. Given the limited window for intervention in many reconstructive procedures, it is widely regarded as unfair to penalize a child for the family's socioeconomic circumstances. Recognizing this, most states have provided health insurance to cover minors for such procedures.

The extent of *distributive justice* is a much more controversial point. To what extent do hospitals or individual physicians owe care to their specific community in contrast to their obligations to the world at large? Many well-meaning surgical missions have pursued cleft care throughout

the world with the belief that all children deserve a chance at normal appearance and speech, whereas some critics decry the steady stream of surgical procedures and therapeutic interventions undertaken on children who cannot fully realize the benefit of their improved appearance because of developmental or other social influences. The wide spectrum of opinion on this issue provides opportunity for debate in a variety of situations.

Regarding justice, there is the additional complication that the surgeon will likely be rewarded financially for operating. Currently, within most American systems, the more surgery one performs, the greater the reimbursement. This provides a definite conflict of interest when parents are looking to the surgeon for guidance on whether to pursue surgery. The ethical surgeon will not knowingly put his or her own financial gain ahead of the best interests of the patient. The hidden costs to the family unit and the perceived benefit to the patient should be carefully considered before a surgical intervention is undertaken.

Once the decision is made to pursue surgery, the medical team must help the patient and his or her family get the most out of that intervention. Certain supportive therapies, such as home nursing, rehabilitative therapy, and even dressing supplies, must be made readily available to all families if they are needed perioperatively. Social work and supportive hospital resources are important parts of serving vulnerable patients. Comprehensive pediatric plastic surgery centers can be especially valuable in bringing together various specialist and supportive therapies in one place to coordinate care and optimize outcomes.

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Within the concept of these guiding principles, the individual principles are seen as binding unless they conflict with other obligations. In situations in which two of these principles are found to be in conflict, such as patient or family autonomy and nonmaleficence, the principles must be weighed and balanced carefully. In the pediatric population, saving the child's life, ensuring quality of life, and nonmaleficence are generally placed in the highest regard among these concerns.

MODERN CHALLENGES

Technology

As modern medicine continues to advance, new challenges arise in the field of medical ethics. In pediatric plastic surgery, many of these new challenges involve our growing understanding of genetics in various syndromes and conditions. Thus knowledge becomes available at an ever-accelerating rate, and the decisions about how to use such information become a trial ground for ethical debates.

Currently, as a result of technology such as prenatal ultrasonography and genetic testing, conditions as varied as complex trisomies, amniotic band syndromes, and fetal cleft lips and palates can be diagnosed just a few weeks or months into gestation. If parents receive one of these diagnoses, there is often the heart-wrenching option to selectively abort the “damaged” fetus to avoid a long string of difficult surgical and therapeutic interventions. No standards exist to say what makes a fetus “undesirable” or “unfit” enough to be born into this world. From a purely financial standpoint, some children are certainly less expensive to society if they were never to come into existence, but this does not address the child's innate value to their family or society. Decisions about the value of human life vary widely among individuals, and the issue remains politically charged. Is it possible that society could face a system someday in which parents are penalized for knowingly bringing resource-demanding children into the world?²¹ Although no one besides the parents in this situation can fully comprehend or make such a difficult decision,

an ethical and careful genetic counselor is invaluable in these situations.²² Likewise, genetic testing should not be undertaken lightly without some advance awareness by the parents that they may be faced with difficult decisions once they receive this knowledge.²²

Legal ramifications complicate the morality of technologic advances when the possibility of both wrongful birth and wrongful life suits against physicians has become a reality. Craniofacial centers have the opportunity to carefully guide parents through the morass of Internet-based information, genetic uncertainty, and the reality of a given genetic condition. As physicians intimately associated with correcting some of these defects, pediatric plastic surgeons are uniquely positioned to assist parents and society in dealing with these issues.²³ In addition, as public policy and legal standards are set, the medical community is certain, and rightfully so, to be called on as advocates for these patients (or these families).

Clinical Research

Another significant ethical dilemma is the role of pediatric patients in clinical research. Surgeons constantly seek to improve outcomes for all patients, and innovation is a key factor in bettering our field as a whole.²⁴ Despite natural hesitations about “experimenting” on children, careful pediatric research, held to the highest ethical standards, is both permissible and to be encouraged. Without research, one cannot improve outcomes, and if medical professionals fail to pursue advances in medicine, they betray their duty to their patients’ very best interests.^{25,26} Similarly, training of future surgeons must continue within the pediatric population to produce well-trained, clinically competent pediatric plastic surgeons.

Media

Media involvement in all aspects of plastic surgery pervades societal expectations of our field. Originally, plastic surgery evolved as a specialty for the treatment of severe mutilating deformities requiring complex reconstruction after World War I. However, in recent years, because of the distorted portrayals of reality television and an increasingly pervasive celebrity culture, a plastic surgery procedure is sometimes viewed as little more than a commodity—much like a facial or fancy haircut. Popular culture has made it seem as though plastic surgical interventions are both instantaneous, without pain or recovery time, and yield flawless results (as presented with the augmentations of expert makeup and skillful lighting). Television programming of dubious quality, such as public “makeovers,” in which women are offered a complex beauty regimen, including multiple plastic surgery procedures, and then made to compete in a beauty pageant, have served to lower the public perception of the professionalism of plastic surgery.

Regarding pediatric plastic surgery, the media is slightly less intrusive. However, even documentary programs on serious conditions that require reconstructive surgery can be misleading as a result of the careful editing and ultimate results that are shown. Patients are seen to zip from life-threatening deformities to a near-normal appearance in the course of an hour, skipping over the logic of careful medical decisions for the drama of “a good television story” and possibly creating unrealistic expectations for patients and their families.

Although the extreme feature of plastic surgery programming has been toned down somewhat in recent years, the constant barrage of society’s fascination with plastic surgery in the media, both celebrated and decried, continues to pervade popular culture. Patients and their families may come to the doctor’s office with preconceived notions from television and extensive Internet searches about what to expect with surgery. In one study of first-time patients seeking a cosmetic surgery consultation, more than half of the patients considered themselves high-intensity plastic surgery television viewers, most of whom acknowledged that these programs influenced their

decision to pursue surgery.²⁷ Should the surgeon deviate from the expected script, patients can be startled or dismayed, and this can disrupt the trust between the patient and doctor. Physicians should accept researched questions, no matter how apparently misguided, as an opportunity to educate the patient on the reality of surgical options and procedures.

Plastic surgeons also need to be aware of their own presence on the Internet and other advertising avenues. For example, “before and after” photographs can mislead if a balanced set of results is not shown. This is an area carefully monitored by the American Board of Plastic Surgery. Other practitioners may not have as much scrutiny and regulation in their advertising. Plastic surgeons must trust in the quality of their work and honest marketing to ultimately maintain the trust of the public.²⁸

SPECIFIC ETHICAL CHALLENGES

Questions of ethics in plastic surgery are most interesting in concrete examples, where the very real conflicts between some of the various principles can be fully appreciated. Take for example, the very real example of craniopagus (conjoined head) twins who presented to attempt separation at 2 years of age²⁹ (Fig. 4-4). The goal of separation was to improve both quality and quantity of life (craniopagus twins’ life expectancy is approximately 9 years). However, despite meeting the goal of beneficence, the risks of death or severe disability separating the twins were very real. The chances that both twins would survive the surgery were approximately 33%, according to the surgical team. Therefore passing the principle of nonmaleficence is an obvious gray area. With regard to patient autonomy, at 2 years of age the twins had minimal comprehension of the proposed surgery, and this decision was entirely on their parents. The justice of bringing the twins from abroad to the United States, where a very resource-intensive team became dedicated to the surgery, is another unclear area. In addition, the media covered the case relentlessly, bringing both additional acclaim and pressure to the treatment team. All of the four principles of medical ethics, even when examined closely, did not yield an easy solution.²⁹

Another complex ethical dilemma arises in how to treat intersex children with ambiguous genitalia at birth. The initial urge is to make these children appear more “normal.” A large number of endocrine and genetic disorders may lead to ambiguous genitalia at birth or unexpected development at puberty. Children believed to have been female at birth may ultimately prove to have XY chromosomes, or vice versa. Previously, children with clitoromegaly or micropenis may have been treated by clitorrectomy with the good intentions of the child going on to lead a “normal life” as a girl. However, these operations can leave the patients scarred, insensate, and with a



Fig. 4-4 Craniopagus twins from abroad brought to the United States for possible separation.

sense of being violated. At times, a child can be truly confused if the parents attempt to hide such a diagnosis because they believe their child will be confused and hurt by this information. Although the parents facing these issues try to “do what’s best” for their children, their motivations for their child being “normal” may ultimately conflict with their child’s needs. They have been concerned about teasing and difficulty in childhood and have sacrificed their child’s autonomy to protect them from being different (despite evidence that surgical outcomes are better in older children). Increasingly, intersex individuals have been critical of this approach, and many experts now agree that it is best to wait until the child is able to have a say in their own surgical treatment.¹⁹

Children born with congenital hand anomalies can also be a challenge for parents. Even in simple ulnar polydactyly, parents struggle with the decision whether to have a digit tied off, which can cause prolonged pain and an unaesthetic remnant, or to forego correction of the physical anomaly until the patient is old enough to receive anesthesia and have it definitively removed. With complex syndactyly or thumb duplication, the options are more challenging still: it is not always clearcut which fingers should be separated or removed and when to do so. If a patient has a largely functional though disfigured hand, the parents may push for a “normalizing” surgical intervention that may not be in the best functional interests of the child. Surgery may lead to joint stiffness, insensitivity, and discomfort, actually making the hand less useful after surgery. In this case, clinicians may urge parents to take a cautious approach, allowing the hand to develop and grow before any surgical intervention is contemplated.

Not unique to children are questions of composite tissue allograft transplants of “nonessential” organs, such as hands or faces. This raises additional questions about assuming the risks of lifetime immunosuppression in exchange for possible increased quality of life. The first partial face replant, which proved some of the feasibility of an allografted face transplant, was performed after a traumatic threshing machine injury in a 9-year-old child in northern India.³⁰ As composite tissue allograft and immunosuppression regimens become increasingly sophisticated, this may well become a treatment possibility for children with burns or other traumatic injuries.

Similar questionable areas arise when older children begin asking for cosmetic surgery. Teenage years are difficult for any child, but one who looks slightly different from their peers can feel truly alone or ostracized. At 16 years of age, we allow children in many areas of the world to operate motor vehicles. Should they have the same right to decide to reshape their nose at that time? This is largely a decision for the child, his or her parents, and the surgeon to reach together, because there are no legal stipulations on cosmetic surgery for minors.

This is the case for many issues in pediatric plastic surgery, from the morality of overseas surgical missions to the proper surgical decisions in the care of intersex babies born with indeterminate genitalia. Are the surgeries intended to make the child feel better or to make society feel better about the child? Careful decision-making is certainly needed in each unique case. The complexity of medical ethics makes it impossible to establish a straightforward, algorithmic approach to every situation. As such, professionalism and the ethical code of plastic surgery must remain sufficiently robust to face these challenging dilemmas with appropriate knowledge and care.

KEY POINTS

- Medical ethics began as a field in Ancient Greece nearly 2500 years ago.
- The majority of medical ethics writing centers around the four key principles propounded by Beauchamp and Childress in 1979:
 - Beneficence requires acting in the best interest of the patient.
 - Nonmaleficence requires acting in a way to avoid harming the patient.
 - Autonomy involves respecting the patient's individual wishes and personhood with respect to medical decisions affecting their body.
 - Justice refers to the fair and sensible distribution of limited and finite resources in caring for patients and society at large.
- Prenatal diagnoses and genetic testing present new challenges for parents and significant dilemmas for medical ethicists.
- Cosmetic surgery in minors is a controversial topic and should be judged by individual patient and family circumstances.
- Universal solutions for ethical dilemmas in pediatric plastic surgery are rare. Rather, each unique case must be examined for the special unique considerations that will guide recommendations for care.

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5

Anesthesia for Pediatric Plastic Surgery

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he primary goal of anesthesia for anesthesiologists, surgeons, and patients is to ensure patient comfort and safety. Events that occur during the period of anesthesia involve a complex interaction of anesthetic technique, surgical technique, and co-existing disease. The anesthetic management of a patient begins with the preoperative visit and can extend well into the postoperative period, including intensive care and pain management. This chapter provides information to assist in the management of all phases of anesthesia care for pediatric plastic surgery patients.

Anesthesia is any technique that allows a patient to withstand a noxious event. The goals of anesthesia are threefold: patient safety, patient comfort, and facilitation of the procedure. Anesthetics combine some or all components of sedation, amnesia, analgesia/anesthesia, and muscle relaxation. They are titrated as needed throughout a procedure and are reversed or allowed to wear off at the completion of the procedure.

Plastic and reconstructive surgery for pediatric patients can involve any part of the body; these children may have a variety of conditions related to existing diseases and syndromes. Safe anesthesia care begins with an understanding of pediatric physiology and development.

DEVELOPMENTAL ANATOMY AND PHYSIOLOGY

Cardiac Physiology

Fetal circulation is characterized by parallel circulation (cardiac output contributes to both pulmonary and systemic perfusion simultaneously, allowing the mixing of oxygenated and deoxygenated blood), whereas in neonates the circulation transitions to series circulation (cardiac output

contributes to either pulmonary or systemic perfusion, with minimal admixture). High pulmonary vascular resistance (PVR) and relatively low systemic vascular resistance also characterize fetal circulation. The foramen ovale and ductus arteriosus allow the shunting of blood around the lungs, with less than 10% of combined ventricular output contributing to pulmonary flow.¹ A series of events occurs at birth that changes fetal circulation into neonatal circulation.

During delivery, PVR decreases and systemic vascular resistance increases, significantly increasing pulmonary blood flow. The increase in systemic vascular resistance occurs as a result of decreased placental blood flow. The onset of spontaneous ventilation creates two factors that cause a precipitous decrease in PVR: a decrease in the mechanical compression of the alveoli and an increase in the partial pressure of oxygen.^{2,3} The increase in partial pressure of oxygen also closes the ductus arteriosus. The changes in pulmonary vascular tone are mediated by biochemical factors, including nitric oxide and prostaglandin. When the PVR decreases, pulmonary blood flow and venous return to the left atrium increase. The increase in left atrial pressure and flow closes the foramen ovale. Over the next few months PVR decreases even further, and the functional closure of the ductus arteriosus and foramen ovale becomes essentially permanent.

In the newborn period, the highly reactive tone of the pulmonary vessels persists. Hypoxemia and tissue acidosis are two important factors that affect PVR. An increase in PVR can lead to right-to-left shunting across the foramen ovale and ductus arteriosus. The persistence of an elevated PVR can lead to further hypoxemia and tissue acidosis. Thus hypoxemia and acidosis can lead to a vicious cycle of increased PVR.

The neonatal myocardium is immature at birth and continues developing postnatally. Many functional differences between the neonatal and adult myocardia are directly related to the immaturity of the neonatal tissue components.⁴ The fetal myocardium has a limited ability to generate the equivalent contractile force, compared with the adult myocardium. The consequence is a reduced capacity to adapt to increases in preload or afterload.^{5,6} Because the neonatal heart has a limited ability to increase stroke volume,⁷ neonates and young infants are poorly tolerant of bradycardia; moreover, significant volume loads may more easily cause ventricular overload and failure.

Respiratory Physiology

A significant difference between neonatal and adult respiration is oxygen consumption. Neonatal oxygen consumption is two to three times greater than that in adults (5 to 8 ml/kg/min versus 2 to 3 ml/kg/min).⁸ This partly accounts for the rapid oxygen desaturation during apnea or hypoventilation.

Neonates and infants also have a higher closing capacity (closing volume plus residual volume) than adolescents and adults. Closing capacity is the lung volume where the collapse of small airways occurs. Neonates have a more compliant chest wall, and a poorly compliant lung. Neonatal/infant lungs are less compliant than adult lungs. Immature lungs in pediatric patients are characterized by small and poorly developed alveoli with thickened walls and decreased elastin.⁹ Elastin provides elasticity to the lung, without which the airways collapse. Because infants (much like older adults) have less elastin, the closing capacity occurs at a larger lung volume in very young and very old individuals.^{9,10} In infants, airway closure can occur before end exhalation (the closing capacity is larger than the functional residual capacity), resulting in atelectasis and right-to-left transpulmonary shunting (Fig. 5-1). An awake, nonanesthetized infant “brakes” exhalation before this point and maintains high respiratory rates to create a dynamic functional residual capacity that is approximately 40% of the total lung capacity, similar to adult values.¹¹ Impairment of this dynamic maintenance of functional residual capacity by anesthesia leads to airway closure, atelectasis, hypoxemia, and right-to-left shunting.

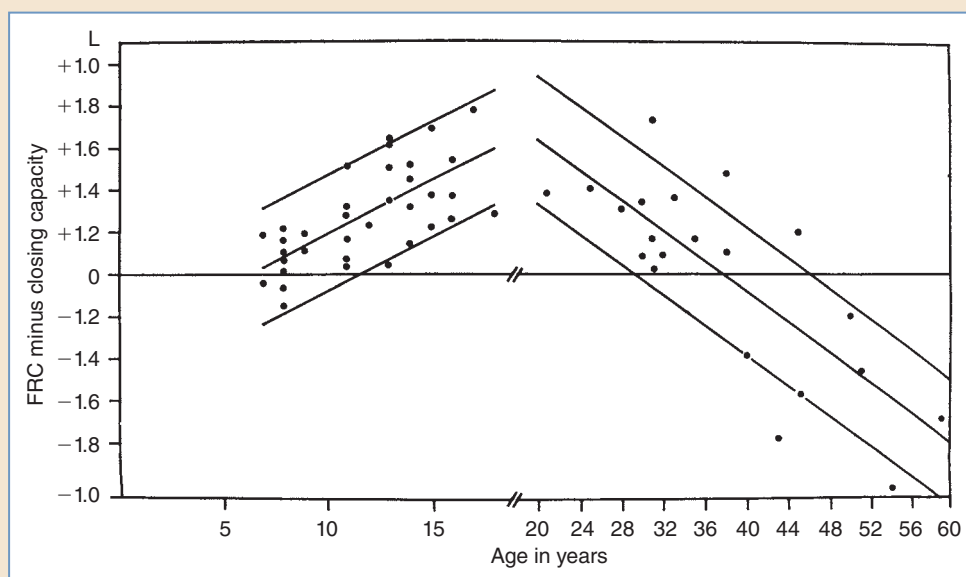


Fig. 5-1 Closing capacity as it relates to age. In children younger than 5 years of age and adults older than 45 years of age, the closing capacity is greater than the functional residual capacity (FRC).

Temperature Regulation

Neonates and infants are at increased risk for thermoregulatory instability, because they are at increased risk for heat loss, and they have a decreased ability to produce heat. They are at increased risk for heat loss because of their large surface area/volume ratio.¹² They also have less ability than adults to restrict heat loss because of limited vasoconstriction.¹³ The primary method of heat production in neonates and infants consists of *nonshivering thermogenesis*. Nonshivering thermogenesis occurs primarily in brown fat, which may be decreased in premature neonates, and compensates poorly for heat loss. This mechanism can also be inhibited by inhalational agents.^{14,15} Shivering thermogenesis is inhibited during anesthesia, because the hypothalamic set point is altered.¹⁶ In addition, the vasodilatory effects of volatile anesthetics can decrease core body temperature by causing admixture of cooler peripheral blood with warm core blood and by increasing passive heat loss to the surrounding environment.

Temperature stability can be ensured by decreasing heat loss to a patient's surroundings and by active warming. The initial step is to increase the ambient air temperature of the operating room; this slows heat loss through convection, conduction, radiation, and evaporation. Radiant warming lights take advantage of the large surface/mass ratio of pediatric patients, and allow access to the patient. Blankets and drapes prevent passive convective heat loss; plastic drapes also prevent evaporative heat loss and can be used in conjunction with radiant heat. Forced warm air blankets and heated water blankets can be used intraoperatively and provide very effective heating. Finally, warming intravenous fluids and blood products may be necessary in some small infants.

Nervous System

There is growing evidence that exposure to anesthetic agents that include volatile anesthetics, ketamine, and benzodiazepines results in accelerated apoptosis (neurotoxicity) in the developing

brain.¹⁷⁻¹⁹ In an animal model, exposure to these anesthetic agents resulted in histologic evidence of apoptosis and cognitive deficits.²⁰ These effects occur during a critical period of brain development, and they appear to be dose dependent. The evidence is not as clear in human studies. Large human retrospective database studies demonstrated an association between general anesthesia and learning deficits. Specifically, an association was noted between the number of anesthetics given in patients younger than 4 years of age and learning deficits.²¹ However, twin studies have not demonstrated these problems in learning or in social behavior.^{22,23} A prospective randomized, multicenter trial (GAS Study) showed that 1 hour of sevoflurane in infants having hernia repair did not cause neurocognitive deficits.²⁴ It is still too early to state how much and at what age anesthesia is completely safe. As more data are produced, guidelines and protocols will likely be developed. It may be premature to limit important surgery for children less than 4 years of age because of fear of deleterious neurocognitive effect.

PREOPERATIVE EVALUATION

Performing a thorough history and physical examination, along with judicious use of laboratory evaluations and other tests, can identify and prevent most potential perioperative problems. Coexisting diseases are common in many pediatric patients undergoing surgery and can complicate surgical and anesthetic management. This section is not a comprehensive review of the perioperative management of coexisting disease but attempts to focus on unique aspects as they relate to anesthesia and pediatric plastic surgery patients.

Prematurity

Prematurity is defined as birth before 37 weeks' gestation. It affects all of the organ systems, and the degree of prematurity is a predictor of mortality.²⁵ The immaturity of the respiratory centers is a major cause of morbidity that can extend beyond the neonatal period. Coexisting diseases of prematurity include bronchopulmonary dysplasia, cognitive and sensory impairments, retinopathy of prematurity, and cerebral palsy.

Premature birth, with or without the potential for ventilator-associated injury, is disruptive to the normal development of the lungs. The use of artificial surfactant, prenatal steroids, and lung-protective ventilation strategies ameliorates this effect. Even after weaning from ventilator support, these children can have lifelong decreases in pulmonary function, compared with their age-matched peers.²⁶ This leaves them vulnerable to increased risks of pulmonary complications under anesthesia. This is very important, particularly in young children, in whom respiratory complications of anesthesia are more common.

Cerebral palsy, characterized by motor spasticity, is common in children born prematurely.²⁵ This may require medical treatment (baclofen) or surgical treatment for contractures. Positioning of patients with contractures is very important to prevent intraoperative pressure injuries. These patients may have swallowing disorders and silent aspiration. Seizure disorders can occur in children born prematurely²⁵ and can be treated medically, or in extreme cases, with vagal nerve stimulators.

Sensory and cognitive impairment is common. This can include retinopathy of prematurity, cortical blindness, strabismus, and hearing impairment. Cognitive impairment ranges from severe, affecting all aspects of functioning, to very subtle forms mostly manifesting as attention-deficit disorders or mild problems in school.²⁵

Apnea of Prematurity

Neonates born prematurely who are less than 45 to 60 weeks of postconceptual age (PCA, calculated by the gestational age plus the age of the infant in weeks) may have episodes of apnea severe enough to cause hypoxia and bradycardia.²⁷ Apnea can be precipitated or exacerbated by anesthesia, which is a concern for all prematurely born neonates who are exposed to sedation or general anesthesia before 45 to 60 weeks' PCA.²⁸ The concern that an undetected, untreated apneic event could lead to postoperative cardiorespiratory arrest must be balanced with the need for the patient to eventually return home. The lower the PCA, the greater the risk for postoperative apnea and the longer the duration of disordered breathing postoperatively.²⁹

Other risk factors include anemia (hematocrit lower than 30%) and a history of previous apnea.^{30,31} It is not clear at what PCA a full-term or prematurely born infant ceases to require postoperative monitoring for these events. In general, a PCA of greater than 45 to 50 weeks may qualify an otherwise healthy prematurely born infant to return home the day of surgery. Children's hospitals have predetermined ages at which a prematurely born infant undergoing general anesthesia must be admitted overnight for observation. Our policy at Children's Hospital of Pittsburgh is to admit all prematurely born infants less than 45 weeks' PCA for apnea and bradycardia monitoring. Infants considered at risk for postoperative apnea should be monitored for at least 12 hours, with an additional 12 hours after any apneic episodes.³¹

Neurology

Neurologic abnormalities can alter the anesthetic management of pediatric plastic surgery patients. Many craniofacial abnormalities are associated with increased intracranial pressure (ICP). The characteristic signs and symptoms of increased ICP include headache, irritability, vomiting, papilledema, and hypertension. Bradycardia with hypertension (Cushing's triad when associated with increased ICP) is a late finding and may signal impending herniation. Many of the symptoms of increased ICP may not be present in children with chronically elevated ICP. Papilledema or abnormal visual-evoked potentials may be the only finding on examination.

Patients with increased ICP present several challenges for anesthesiologists. The anesthetic goal is to maintain cerebral perfusion pressure by maintaining mean arterial pressure and minimizing ICP. The risk of aspiration is also higher in patients with acutely elevated ICP and presents a challenge, particularly in patients with a difficult airway. Patients with acutely elevated ICP are typically anesthetized with a rapid-sequence induction using an induction agent that maintains mean arterial pressure and decreases ICP, such as sodium pentothal or etomidate. All of the volatile inhaled anesthetic agents increase ICP in a dose-dependent manner by increasing cerebral blood flow. This has been demonstrated in children even at low concentrations (half the minimum alveolar concentration).³² Techniques to acutely lower ICP include the following:

- Elevating the head of the bed
- Hyperventilation
- Administering barbiturates
- Administering osmotic diuretics (mannitol [Osmitrol]) and loop diuretics (furosemide [Lasix, Lo-Aqua])
- Administering sedation
- Draining cerebrospinal fluid

Obstructive Sleep Apnea

Obstructive sleep apnea (OSA) or sleep-disordered breathing is caused by periods of upper airway collapse during normal somatic muscle relaxation during sleep. Frequent and/or prolonged periods of airway collapse can lead to disordered sleep architecture, hypoxia, and hypercarbia. Daytime manifestations in children may not include sleepiness or morning headache, but instead may manifest as irritability and developmental delay. Systemic hypertension, and even pulmonary hypertension, can occur with prolonged exposure to hypoxic events.

Many craniofacial abnormalities and syndromes may be associated with OSA. Tonsillar hypertrophy is one of the most common causes of OSA in children. Obesity is also associated with OSA, and many otherwise “healthy” obese children have undiagnosed sleep apnea. Mechanical factors contributing to sleep-disordered breathing in obese patients include upper airway obstruction, restrictive chest excursion, increased work of breathing, and decreased FRC secondary to adipose tissue. Infants and children with craniofacial anomalies, including micrognathia and midface hypoplasia, can have significant OSA.

The central causes of sleep-disordered breathing occur in patients with prematurity or neurologic abnormalities such as cerebral palsy. Chronic hypoxia/hypercarbia from OSA can lead to central apnea from a reduced respiratory drive and can even increase sensitivity to opioids.^{33,34}

Patients with OSA and sleep-disordered breathing can have disordered breathing while recovering from the sedative effects of anesthesia.^{35,36} Except for the most superficial procedures, patients with severe OSA may be poor candidates for same-day surgery. The intensity of post-operative monitoring depends on coexisting conditions, the severity of OSA/sleep-disordered breathing, and the nature of the operative procedure.

Upper Respiratory Tract Infection

Signs and symptoms of an upper respiratory tract infection may be the most common reason for canceling elective surgery.³⁷ Patients with symptoms of a current or recent upper respiratory tract infection are more prone to developing adverse respiratory events such as laryngospasm, bronchospasm, and oxygen desaturation. These risks are higher with decreasing age, baseline reactive airway disease, prematurity, parental smoking, and surgical or anesthetic manipulation of the airway.³⁷ These children may benefit from having their surgeries postponed for 2 to 6 weeks. The decision to cancel surgery for a child with an upper respiratory tract infection must take into account the urgency of the surgery, the medical condition of the child, and the likelihood that the child will return asymptomatic if the surgery is rescheduled. An otherwise healthy child who is scheduled for a superficial procedure may do quite well with an upper respiratory infection, unlike a child with a coexisting respiratory disease who requires airway manipulation as part of the procedure.

Congenital Heart Disease

Cardiac lesions may accompany craniofacial anomalies. In addition, many nonsyndromic children who are seen for plastic surgery can have coexisting congenital or acquired cardiac disease. As surgical treatment of complex congenital cardiac disease has improved, more patients are now seen with partially or fully palliated lesions.

Initial assessment includes information regarding functional status. In otherwise healthy children with no history of physical limitations (normal bottle feeding or ability to keep up with peers) and normal physical examination results, no further testing is required. In patients with severe or complex cardiac disease, preoperative assessment may require coordinating with the

patient's cardiologist to ensure an uneventful perioperative course. Recent data from electrocardiograms, echocardiograms, and catheterization can provide up-to-date information about rhythm abnormalities, ventricular function, the type and severity of valvular lesions, and the presence and patency of conduits. A complete set of vital signs, including baseline oxygen saturation, should be recorded. This information helps to define operative risk and a plan for intraoperative and postoperative management. Prophylaxis for subacute bacterial endocarditis should be provided as per the American Heart Association guidelines.³⁸

Malignant Hyperthermia

Malignant hyperthermia is triggered by volatile anesthetics and the depolarizing neuromuscular blocker succinylcholine (Anectine and Quelicin). Exposure to these substances causes susceptible patients to have uncontrolled metabolism of muscle energy stores, leading to hypercapnia, tachycardia, hyperthermia, rhabdomyolysis, and death if untreated. Susceptibility to malignant hyperthermia is a genetic disease showing autosomal dominant inheritance in most cases. Some centers use muscle biopsy and halothane-caffeine contracture testing to determine whether older patients or family members are susceptible.³⁹ Unexplained incidents during anesthesia, especially when accompanied by high fever, should be explored both in patients and their family members; when in doubt, a nontriggering anesthetic (devoid of volatile agents and succinylcholine) should be used.

Airway Examination

A thorough examination of the airway may be difficult with extremely young or otherwise uncooperative patients, but it is important for anticipating airway management challenges before they occur. Associated anomalies that can present a challenge include facial and airway features that make mask ventilation and intubation difficult. Features that may predict difficulty with mask ventilation include midface hypoplasia and an enlarged tongue. In addition, a small mandibular space, small jaw opening, decreased translocation, and decreased neck range of motion can be predictive of difficult intubation. Cervical spine fusion, which may occur with Apert syndrome, can make endotracheal intubation even more challenging if neck mobility is decreased. These features may improve after surgical reconstruction, or they may be exacerbated during normal growth of the involved structures or because of scarring.

Laboratory Testing

The yield of preoperative screening tests in otherwise healthy children is low.⁴⁰⁻⁴³ Testing should be focused on assessing the baseline of a preexisting abnormality, particularly if it may be affected by the surgical procedure. For reconstructive surgeries that involve significant blood loss, the child's preoperative hematocrit level and blood type and crossmatch should be determined. In addition, clotting studies should be performed for surgical procedures that involve significant bleeding. These may include a preoperative prothrombin time, partial thromboplastin time, and platelet count. Some centers also perform dynamic clotting studies such as closure time (platelet function) or a thromboelastogram. Patients with OSA may have polysomnography performed preoperatively. The apnea-hypopnea index (AHI) describes the number of apneic and hypopneic events during an hour of sleep. The severity of OSA by this index is shown in Table 5-1. These ranges are different for infants and children than for adults. A range of 0 to 4 may be normal in adults, and a range of up to 20 is considered mild OSA. This is not the case for children.⁴⁴

Table 5-1 Severity of Sleep Apnea in Children

AHI	Severity of OSA
1-5	Mild
6-10	Moderate
>10	Severe

AHI, Apnea-hypopnea index; OSA, obstructive sleep apnea.

Table 5-2 Fasting Guidelines

Substance	Fast Time (hours)
Clear liquids*	2
Breast milk	4
Formula	6
Nonhuman milk†	8

*Clear liquids include fruit juice without pulp, tea (without milk), black coffee, and water.

†Six-hour fast in infants younger than 6 months of age.

Fasting Guidelines

The absence of airway reflexes during general anesthesia places patients at risk for passive regurgitation of stomach contents and subsequent introduction into the lungs. This can lead to aspiration pneumonia from contact with acidic gastric contents, and possible pneumonia. Risk factors for aspiration include patients being seen on an emergency basis (these patients are more likely to have large volumes of gastric contents) and patients with high disease severity.^{45,46}

Historically, assurance of an “empty stomach” relied on patients being placed on nothing by mouth after midnight the day of their procedure. This can mean periods of fasting from 8 to 16 hours or more, depending on the time of surgery. In addition to being unpleasant, prolonged fasting in patients without access to intravenous fluids or glucose can lead to dehydration and hypoglycemia, especially in very young patients. Liberalization of NPO guidelines (Table 5-2) has increased patient and parental satisfaction without increasing residual gastric volume or aspiration rates.⁴⁷ Exceptions may include patients with reflux or inflammatory bowel disease, who may have normal fluid transit but abnormal solid transit. In particular, solids follow zero-order kinetics: Even small amounts of solid food may pose a significant aspiration risk.⁴⁸

ANESTHETIC MANAGEMENT

Premedication

The prospect of a surgical procedure can produce anxiety for patients of all ages. Going into the unfamiliar environment of an operating room provokes fear of the unknown (“What is going to happen to me? Will it hurt?”) and the known (postoperative pain, needles, and induction). A

Table 5-3 Sedative Premedications

Drug Name	Route and Dose	Benefits and Disadvantages
Midazolam*	PO: 0.5-1 mg/kg IV: 0.05-0.1 mg/kg IM 0.1-0.2 mg/kg Nasal: 0.2 mg/kg	Benefits: Excellent anxiolysis, amnesia, enhances cooperation, onset in 15 min with PO route, 5 min with nasal route Disadvantages: Bitter taste, pain with nasal route, rare paradoxical agitation
Ketamine	PO: 2.5-10 mg/kg IV: 1-2 mg/kg IM: 2-10 mg/kg	Benefits: Rapid onset, excellent sedation, excellent analgesia, mood elevation Disadvantages: Secretions, hallucinations, may lower seizure threshold, rare nausea
Clonidine	PO: 4 µg/kg	Benefits: Antiemetic, analgesic, reduces anesthetic requirement, tasteless, reduces emergence delirium
Dexmedetomidine	PO: 2 µg/g Nasal: 1 µg/kg	Disadvantages: Prolonged onset time (40 min), minimal anxiolytic effect

*Ketamine, clonidine, or dexmedetomidine can be combined with midazolam for an additive effect. The doses can be reduced by 25% to 50% if combination therapy is used.

IM, Intramuscular; IV, intravenous; PO, per os.

special consideration for pediatric patients is separation from their parents or caregivers and application of a mask for inhalation induction of anesthesia. Separation anxiety begins to develop at approximately 7 or 8 months of age.⁴⁹ Separation from a familiar caregiver and introducing a face mask can cause tremendous anxiety and could contribute to increased emergence delirium, increased postoperative pain, and postoperative behavioral changes such as bed-wetting, increased separation anxiety, and nightmares. These postoperative behavioral changes are typically short lived, usually lasting no more than 2 weeks.⁵⁰

One approach for minimizing this trauma is to give a sedative medication before patients are separated from their parents. The goal of this technique is to produce calm, cooperative children who can be taken to the operating room and have an uneventful induction of general anesthesia. The use of a sedative premedication can produce a less anxious induction of anesthesia and may also lower the chances of negative postoperative behavioral changes.⁵⁰ In patients with coexisting diseases, it may be important to balance the risks of an uncooperative child with the risk that sedation may cause airway embarrassment (obstruction resulting from structural abnormalities, bleeding, or foreign bodies) or increased ICP. Other medications used for sedative premedication include ketamine and alpha-2 agonists such as clonidine or dexmedetomidine. These medications are described in Table 5-3.

Another strategy for minimizing separation anxiety and enhancing cooperation is to have a parent present during the induction of anesthesia.^{51,52} If handled correctly, parental presence may be as effective as premedication with midazolam for preventing separation anxiety. However, it is not as effective as midazolam in reducing anxiety associated with the introduction of a face mask.⁵¹ Parental satisfaction is very high when a parent is present during the induction of anesthesia, and most parents would choose to repeat the process in the future.⁵³ However, children who undergo induction of anesthesia in the presence of extremely anxious parents may actually have more anxiety and more negative behavioral changes postoperatively than if anesthesia had been induced in the standard manner.⁵⁴

Monitors

The American Society of Anesthesiologists' standard anesthetic monitors include continuous ECG recording, noninvasive monitoring of blood pressure, and continuous monitoring of oxygenation with pulse oximetry. Continuous end-tidal carbon dioxide monitoring is required if an airway device is used. If a ventilator is in use, a device capable of sounding an audible alarm in the event of an accidental disconnection must be in place. Temperature monitoring must be available to monitor and/or correct intraoperative changes if necessary.⁵⁵

Depending on the surgical procedure and the patient, other monitoring may be necessary. Catheterization of the bladder can provide information regarding volume status, hemoglobinuria, or myoglobinuria and can decompress the bladder during lengthy procedures. Measuring arterial blood pressure may be necessary for patients requiring frequent intraoperative blood sampling (surgeries with significant blood loss), for patients with preexisting or acquired hemodynamic instability requiring beat-to-beat blood pressure assessment, and for patients for whom noninvasive blood pressure measurement is unreliable (such as morbidly obese patients). Central venous pressure monitoring may be necessary for patients in whom vascular access is difficult, when urine output is not a reliable measure of volume status because of renal dysfunction, or during procedures with significant volume shifts (such as third-spacing or bleeding) for which central venous pressure trends may be helpful.

Monitored Anesthesia Care

The definition of *sedation* varies, depending on the specialty. For patients and families, sedation implies anxiolysis and analgesia, with preserved awareness; this may be undesirable for many patients for whom unawareness of the procedure is paramount. For medical personnel, sedation implies minimal perturbation of respiratory drive and airway reflexes, allowing perhaps less hemodynamic and respiratory complications and therefore decreased use of medical resources and faster recovery. In practice, this all depends on the patient, the agents used, and the procedure for which sedation is necessary. All agents have the potential to decrease airway reflexes, despite the preservation of consciousness. The deeper the level of sedation, the greater the possibility of depressed airway reflexes and respiratory and hemodynamic depression. Ideally, the procedure planned should be painless either from the beginning or with judicious application of local anesthesia. In practice, sedation frequently results in a patient who is unresponsive to noxious stimuli. Any sedation procedure should prepare for the possibility of respiratory depression and airway obstruction. The risk of this transition increases when the procedure requires patient immobility, especially with greater levels of noxious stimuli.⁵⁶ Equipment and trained personnel must be available to rescue a patient from the next level of sedation.

General Anesthesia

General anesthesia has the following four characteristics: unconsciousness (asleep), immobilization (akinesis), no pain (anesthesia), and no memory (amnesia). The induction of general anesthesia is the first step in the transition from an awake state to the anesthetized state. This can be accomplished through an inhalation, intramuscular, or intravenous technique. During the induction of general anesthesia, airway management is necessary to prevent hypoxia.

Inhalation (or mask) induction is common for pediatric patients. After the monitors are placed (or during placement with uncooperative patients), a mask connected to the anesthesia machine is placed on the patient, and a mixture of nitrous oxide, oxygen, and a volatile agent (sevoflurane) is given. The inhaled anesthetic agents are delivered to the brain after they are dissolved in the

blood, passing through the alveoli. Many patients become quite claustrophobic from placement of the mask and dislike the pungent odor of the volatile agents. Gentle introduction, scenting of the mask with flavoring, and initiation of odorless nitrous oxide before the introduction of a volatile agent may make this technique less unpleasant.

As a patient inhales the anesthetic agents, an increasing amount becomes dissolved in the blood; this action resembles the administration of a continuous intravenous infusion. The patient transitions from a conscious state to one of excitement and disinhibition, commonly referred to as stage 2. This stage is characterized by increased airway reactivity, and the patient may go into bronchospasm or laryngospasm in this stage with minimal stimulation. If a patient regurgitates stomach contents during this stage, aspiration can occur. After the child is completely anesthetized, noxious stimuli such as placement of an intravenous catheter are well tolerated.

The advantages of inhalation induction are the initial avoidance of needles and the potential for preserving respiration. After a state of general anesthesia is achieved, intravenous access is facilitated both by patient immobility and by the vasodilatory effects of volatile anesthetics. Moreover, it is easier to maintain a difficult airway when a patient continues to breathe spontaneously. Unfortunately, this technique does not prevent regurgitation and aspiration of stomach contents; therefore it is contraindicated in patients with a full stomach. Patients with a mask phobia may not tolerate it well and may prefer the discomfort of a needle rather than have a mask placed on their face.

Rapid induction of general anesthesia can be achieved using intravenous administration of anesthetic agents. For elective procedures, most children and many adolescents fear intravenous injection more than the procedure itself. Reassurance, premedication, preoperative topical anesthetics such as a eutectic mixture of local anesthetics (EMLA), and nitrous oxide may make venipuncture less stressful and painful. The rapid onset of general anesthesia is particularly advantageous for patients with a full stomach who require a surgical procedure, because a secure airway can be established before regurgitation of stomach contents becomes likely; this is known as *rapid sequence induction* (RSI). In addition, intravenous induction of anesthesia is odorless and does not induce claustrophobia. Mask inductions can be difficult with adolescents, because these patients can be uninhibited, strong, and combative during induction.

Airway Management

There are several goals of airway management during general anesthesia. The primary goal is the provision of a patent conduit to the lungs; the airway may become obstructed during unconsciousness, or it may become blocked with stomach contents, blood, or secretions. Another goal is the support of ventilation; hypoventilation and apnea can occur during general anesthesia, and controlled ventilation may be necessary. A third goal is delivery of oxygen and/or anesthetic agents instead of room air. Finally, any airway management strategy must facilitate the surgical procedure being performed.

Natural Airway

The natural airway strategy is appropriate only if the patient does not have a full stomach and does not require positive pressure to maintain a patent airway and adequate ventilation. Nasal or oral airways may assist the patient in maintaining a patent airway with spontaneous ventilation. Supplemental oxygen can be administered, but delivery of gaseous anesthetic agents is unreliable. Infusion of intravenous agents can maintain general anesthesia.

Mask

With a mask, ventilation can be spontaneous or partially or completely controlled. A patent airway can be achieved with gentle positive pressure support in patients who are breathing spontaneously,

or it can be assisted with oral or nasal airways. A tight mask seal allows reliable administration of oxygen and gaseous anesthetic agents. For quick procedures around the face or mouth (such as arch bar removal), a mask can be removed briefly and replaced periodically to deliver more anesthetic. Because the airway is not manipulated, this is the ideal technique for patients with reactive airway disease. However, it does not protect against laryngospasm or regurgitation.

Laryngeal Mask Airway

A laryngeal mask airway (LMA) involves a strategy similar to that used with a normal mask and does not enter the glottis but instead directs air into it. Because it is seated in the oropharynx, past the tongue, it can mechanically create a patent airway in patients with collapse of soft tissues in this area during anesthesia. A proper seal is created that prevents introduction of room air and provides a very reliable delivery of gaseous anesthetics. The patient may be allowed to breathe spontaneously, or ventilation may be controlled. Because of the greater ease of achieving a tight seal and the position of the LMA beyond the soft tissue of the oropharynx, lower peak pressures may be needed to deliver a positive pressure breath than with mask ventilation. Indeed, the mask airway can be used as an emergency airway device for patients who are difficult to ventilate using a mask and in whom intubation is initially unsuccessful.⁵⁶

Once an LMA is placed, ventilation can continue even at a distance from the anesthesiologist. In addition, because less of the face is covered than with mask ventilation, it can be used during procedures for which mask ventilation would likely interfere, such as removal of facial or nasal nevi or dermoid cysts. As with a mask, the airway is minimally manipulated; thus the risk of bronchospasm is lower in patients with reactive airway disease. However, similar to a mask, it does not protect against laryngospasm or regurgitation and is contraindicated in patients with a full stomach.

Endotracheal Tube

An endotracheal tube provides definitive airway management in unconscious patients. It directly enters the trachea, preventing airway obstruction from laryngospasm and minimizing airway soiling. High positive pressures can be delivered with a cuffed endotracheal tube or with an uncuffed tube of appropriate size. Direct stimulation of the trachea can precipitate coughing and bronchospasm under light anesthesia, especially in patients with reactive airway disease. In addition, patients may develop edema at the site of the tube, causing stridor or croup after extubation. An appropriately sized endotracheal tube minimizes this risk. Assessing the degree of air leakage around the tube can also help decrease the risk of tracheal edema and postextubation croup. In general, a leak that is generated at pressures at or below 25 cm H₂O will not cause significant tracheal edema.

Endotracheal tubes come in many sizes and shapes. They have the lowest profile of any airway device and can be placed in the trachea using the nasal or oral route. Placing an endotracheal tube accurately in the trachea usually requires a direct or indirect view of the glottis and surrounding structures. Two exceptions are blind nasal intubation, in which the tube is advanced nasally until breath sounds are heard clearly through it, and the use of a lighted stylet, which illuminates the tip of the endotracheal tube through the sternal notch when it is at the glottis. If any doubt exists about whether a patient can be intubated using a direct view, an alternative technique that is familiar to the anesthesiologist must be immediately available.

Using a direct view, the tube can be seen passing into the larynx looking directly through the patient's mouth. A laryngoscope blade of the appropriate size and shape is used to align the oropharyngeal-laryngeal axis, and the tube can thus be placed through the mouth or nasopharynx directly through the glottis. In some patients this alignment may not be possible because of unfavorable anatomy. At this point the tube can be passed blindly into the glottis if any part

of the larynx is visible as a landmark (posterior arytenoids or epiglottis), or the attempt can be abandoned and another technique used.

Patients with craniofacial anomalies require anesthesia for a variety of procedures, including direct laryngoscopy, tongue lip adhesions, cranial vault reconstructions, and distraction osteogenesis. Some of these patients may have difficult airways, and the anesthesiologist may encounter airway obstruction and a difficult intubation. This can result in intraoperative and postoperative respiratory complications.

Intubation for patients having plastic surgery is performed using a variety of techniques. Some patients may require a nasal intubation (tongue-lip adhesion), but most will need oral intubation. If a patient has been identified as having a difficult airway, specific techniques and equipment may be helpful. The hallmark for managing a known pediatric difficult airway is to maintain spontaneous ventilation. Several airway techniques have been described to assist. These include LMAs (including the air-Q devices),^{57,58} fiberoptic scopes, the GlideScope,⁵⁹ the Shikani scope,⁶⁰ and Airtraq devices.⁶¹ General anesthesia may be induced before securing the airway; however, maintaining spontaneous ventilation is essential. This can be very difficult, and the anesthesia care provider may need to use airway adjuncts such as a two-handed jaw thrust, oropharyngeal airway, nasopharyngeal airway, and LMA. Laryngoscopy should be performed for intubation if the airway remains obstructed despite these maneuvers. If intubation still is not possible, an emergent bronchoscopy with a rigid bronchoscope, performed by Otolaryngology, may secure the airway. An emergent tracheostomy may be considered but is not ideal as an initial rescue maneuver in a pediatric “cannot intubate, cannot ventilate” scenario, because it is time consuming and technically challenging. Extracorporeal membrane oxygenation may be an alternative rescue option in centers where this resource is available.

A paraglossal approach may be more effective than standard laryngoscopy when intubating patients with micrognathia. This technique has been described in patients with Pierre Robin sequence.⁶² It involves placing the laryngoscope in the right corner of the mouth and passing it along the groove between the tongue and the tonsils, using leftward and anterior pressure. The tongue is displaced to the left, and no part of the tongue hangs over the right side of the blade at any time. This shortens the distance to the glottic opening⁶³ (Fig. 5-2).

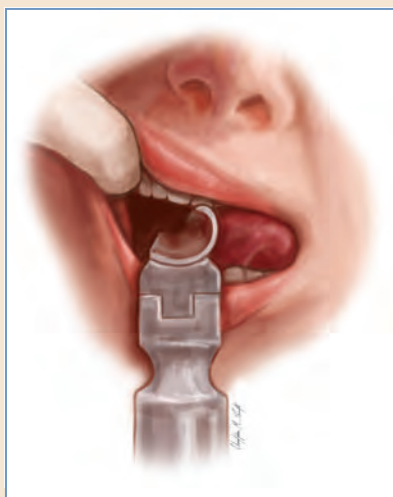


Fig. 5-2 Paraglossal approach for direct laryngoscopy. The tongue is swept completely to the left.

Other techniques can facilitate intubation in pediatric patients, and they may prove to be useful in pediatric plastic surgery patients. These include the use of flexible fiberoptic scopes, video laryngoscopes (Storz videolaryngoscope, GlideScope), and ultrasonography. A fiberoptic scope can be placed orally through an LMA or nasally to visualize the glottic opening. Nasally, a nasopharyngeal airway can be placed in the opposite naris and used as a conduit to introduce a volatile anesthetic agent and oxygen into the posterior pharynx of the spontaneously breathing patient. The Storz video laryngoscope helped to facilitate intubation in a 9 kg infant with Pierre Robin sequence.⁶⁴ Fladjoe et al⁶⁵ recently compared the GlideScope with direct laryngoscopy in infants and neonates. Although none of these patients had craniofacial anomalies, the GlideScope was effective in patients younger than 1 year of age. The time to obtain the best laryngoscopic view was faster with the GlideScope, compared with direct laryngoscopy with a Miller No. 1 blade. However, the time to pass an endotracheal tube was slower with the GlideScope. The GlideScope has different disposable adapters and per the manufacturer may be used in neonates as small as 1.5 kg.

Patient Positioning

After induction of anesthesia, a patient may need to be repositioned for the procedure. Bony prominences and limbs should be padded and positioned to prevent undue tension and pressure on joints and major nerves. Delicate structures such as eyes, ears, genitals, and fingers should be checked and protected. Patients with proptosis may develop eye injuries resulting from corneal abrasions and globe pressure. The eyes need to be closed or well lubricated, and no pressure should be placed on the globe, especially for patients placed in the prone position.

The airway is a particular concern; it can be dislodged during repositioning and at any time during the procedure if undue tension develops along the anesthesia circuit. Careful taping and positioning of the endotracheal tube and anesthesia circuit decreases but does not eliminate the risk of accidental disconnection. Attention to early signs of disconnection (disconnect alarms, end-tidal carbon dioxide) is vital, because some patient positions (prone) may make resumption of ventilation difficult.

Anesthesia Maintenance

Anesthesia is maintained with inhaled anesthetics, intravenous anesthetics, or both. In patients who are inadequately anesthetized, movement can occur that disrupts the procedure, and awareness is also possible. Total muscle relaxation may be necessary to facilitate surgical exposure.

The depth of anesthesia is assessed using several pathways. Observing heart rate and blood pressure responses to noxious stimuli is helpful but lacks sensitivity⁵⁸ and specificity. Monitoring end-tidal concentrations of inhaled anesthetics is a useful adjunct to monitoring vital signs. These concentrations are equivalent to end-organ concentrations at steady state.

Most patients come to the operating room with a fluid deficit from their preoperative fast. In addition, the anesthesiologist must replace third-space loss, blood loss, urine output, and insensible losses intravenously. In patients with normal renal function who are not on diuretics, urine output can be a good indicator of volume status. Former premature infants and infants less than 2 months of age should have their glucose levels monitored, because their metabolic response to fasting may be inadequate to prevent hypoglycemia.

Blood loss can be assessed in the operating room by observing the suction canisters, weighing the sponges, and using portable quantitative measurements of hemoglobin, such as the HemoCue. Transfusion triggers for anesthetized patients are not identical to those for awake patients. Because of their decreased metabolism, anesthetized patients are typically more tolerant of anemia, and severe anemia in anesthetized patients is invariably asymptomatic and often well tolerated.

For many pediatric patients, a hemoglobin of 7 g/dl can function as a transfusion trigger in those who are hemodynamically and physiologically stable with no significant ongoing hemorrhage.⁶⁶

POSTOPERATIVE CONCERNS

Emergence

When the administration of anesthetic agents ceases, the patient clears them through exhalation, metabolism, or redistribution and begins to emerge. Emergence mirrors induction, with the patient passing first through general anesthesia and then excitement before finally regaining consciousness. All cautions and concerns regarding induction also apply to emergence.

As with induction, manipulating the airway when the patient is inadequately anesthetized (stage 2) can precipitate bronchospasm and/or laryngospasm. For a patient under general anesthesia with an empty stomach and a clear pharynx, it is possible to remove any airway devices and support the airway until the patient is able to do so independently. A patient with a full stomach or a potentially difficult airway must regain airway reflexes before extubation. Signs that a patient has regained airway reflexes include eye opening, purposeful movement, and/or cooperation with simple commands (in older patients); coughing, swallowing, and nonpurposeful movement are not adequate signs.

Recovery

Emergence from anesthesia can extend well into the postoperative period; 30% of deaths attributed to an anesthetic occur immediately postoperatively. Many of the concerns associated with patients in the immediate postoperative period involve the airway. Sedation both from the recent anesthetic and from pain medication given in the recovery room can cause hypoventilation and airway obstruction. These may resolve with supplemental oxygen, stimulation and repositioning, and/or placement of a nasal airway but may require pharmacologic reversal of benzodiazepines or opioids and/or reintubation. Airway obstruction from swelling of the airway and the soft tissues surrounding it can present either in the operating room or immediately thereafter in the recovery room, exacerbating the situation. The airway must be secure before a patient leaves the high-intensity monitoring of the recovery room. Those who still require close observation and frequent interventions may need to be transferred to an intensive care unit.

As patients emerge from anesthesia, they may have nausea and vomiting. Postoperative nausea and vomiting (PONV) is the most common immediate postoperative problem with anesthetized children.^{67,68} In addition to possibly causing dehydration, electrolyte derangements, and surgical site damage from persistent or violent episodes, vomiting can be unpleasant for patients and their parents.⁶⁹ With ambulatory patients, PONV is a common reason for prolonged length of stay or unplanned postoperative hospital admission.⁶⁹⁻⁷⁵ The likelihood of a patient having PONV depends on a combination of patient, surgical, and anesthetic factors.

Intrinsic patient factors such as age, sex, and a history of motion sickness cannot be changed, but this information can help identify patients at risk for PONV. Studies examining PONV tend to find the lowest rates in children up to 3 years of age and show the risk of PONV increasing with age.^{67,76-79} Female sex is not a risk factor in prepubescent children but plays a factor after adolescence.^{77,80} A history of previous PONV or motion sickness is also predictive.⁷⁷ Patients who smoke are at lower risk of PONV.^{80,81} It is not yet known whether secondary smoke exposure decreases risk in a similar manner.

Although some studies have not found the type of surgery to be an important factor with PONV,^{71,81} other studies have shown that ear, nose, and throat surgery, and especially strabismus surgery, pose a greater risk.^{68,77,79} This disagreement may occur because some surgical procedures may be more likely to be associated with other emetogenic risk factors such as postoperative administration of opioids, duration of surgery, and endotracheal intubation.

Many common anesthetic drugs affect the risk of PONV. Halogenated anesthetics increase risk, whereas propofol (Diprivan) decreases risk.⁸⁰⁻⁸² The relationship between nitrous oxide and PONV is not as evident. Some studies show a link,^{81,83} but others do not.^{72,84,85} Vigorous hydration (approximately 30 ml/kg) can decrease the risk of PONV.^{86,87} Finally, although postoperative pain itself can precipitate PONV,⁸⁸ postoperative opioid administration is a risk factor for PONV.^{77,80}

Eberhart et al⁸⁹ created a risk stratification to identify pediatric patients most at risk for PONV. The four risk factors identified are the following:

1. Age older than 3 years
2. Surgery longer than 30 minutes
3. A history of PONV in the patient or family (parent or siblings)
4. Strabismus surgery

The presence of each factor increased the risk by 10%, 30%, 55%, and 70%.

Management strategies for preventing and/or managing PONV depend on a patient's risk for PONV and the potential consequences of PONV in that patient. For patients at low risk, a single intervention or expectant management may be appropriate. High-risk patients may require multiple interventions.^{81,90} In particular, the combination of dexamethasone and serotonin antagonists can be very potent against both early and late PONV.^{72,73} For patients whose treatment has been unsuccessful, treatment with a drug in a different modality or class is indicated.⁹⁰

Some patients develop emergence delirium postoperatively. Emergence delirium is common in preschool-age patients and is characterized by confusion, inconsolability, psychomotor excitement, and dysphoria. This may resolve with administration of sedative or analgesic medications, parental presence, or time. This state is distressing to witness and can lead to patient injury (falling, pulling out intravenous lines, drains, and monitors), caregiver injury, and the need to delay discharge.⁹¹ Awakening to pain may be a risk factor; pretreatment with nonsedating analgesics has decreased the incidence in some series.⁹² Emergence delirium can be treated with a short-acting analgesic such as fentanyl (Sublimaze) if pain is thought to be a contributing factor. The use of dexmedetomidine intraoperatively as prophylaxis or as rescue therapy has become common in pediatric anesthesia. A benzodiazepine such as midazolam may also reduce agitation behaviors. Because patients with emergence delirium may be dangerous to themselves and others, they require intensive nursing care and should not leave the recovery area until their condition resolves.

Several checklists and scoring systems are available to evaluate readiness for discharge from the recovery room. Table 5-4 highlights the clinical features of the Aldrete scoring system.⁹³ The maximum score is 10, and a score of 8 or higher indicates readiness for discharge from the recovery room. In general, satisfactory management of the previously mentioned issues should be sufficient for transfer of patients to a lower intensity of care.

One especially disturbing complication of anesthesia is intraoperative awareness. In adults, the incidence is approximately 0.1% to 0.2%.⁹⁴ The pediatric incidence is difficult to assess because of limited data, but it may be up to fourfold higher.⁹⁵ Anesthesiologists and surgeons alike routinely reassure their patients that they will be unconscious during their procedures. When this expectation is not fulfilled, anxiety and distress result. Consequently, intraoperative awareness has been associated with persistent negative psychological sequelae in adults, including case reports of posttraumatic stress disorder.⁹⁶ However, data do not currently indicate any long-term effects of intraoperative awareness in children.⁹⁵ If a patient or family member describes such an experience, it should be frankly discussed and documented in the chart. The use of a bispectral index to decrease the incidence of intraoperative awareness is intriguing⁹⁷; although data in pediatric patients are sparse, the use of these monitors may be appropriate in patients who are at increased risk for intraoperative awareness. A risk factor for awareness in adults appears to be the use of neuromuscular blocking agents. It is not clear that this is true in pediatric patients. A possible risk factor for pediatric patients is the use of induction rooms.⁹⁸

Table 5-4 The Aldrete Score

Criteria	Score
Able to move four extremities voluntarily	2
Able to move two extremities voluntarily	1
Unable to move extremities voluntarily	0
Able to breathe deeply and cough freely	2
Dyspnea or limited breathing	1
Apneic	0
Blood pressure $\pm$ 20% of baseline	2
Blood pressure $\pm$ 20%-49% of baseline	1
Blood pressure $\pm$ 50% of baseline	0
Fully awake	2
Rousable on calling	1
Not rousable	0
Maintains O ₂ saturation >92% on room air	2
Needs O ₂ to maintain saturation >92%	1
O ₂ saturation <90% with supplemental O ₂	0

The maximum score is 10. A score higher than 8 is required for discharge from the recovery room.

Pain Management

Pediatric patients have pain after many of the plastic surgery procedures. Postoperative pain can range from mild (for example, during arch bar removal or examination under anesthesia) to moderate (after a cleft lip or palate repair) to severe (after rib and iliac crest bone grafting). Intraoperative and postoperative pain can be managed with nonopioid analgesics, opioids, and regional anesthesia. Nonopioid analgesics primarily consist of acetaminophen and NSAIDs. Acetaminophen is well tolerated and can be given orally, rectally and intravenously. NSAIDs include ibuprofen and ketorolac. Ketorolac can be used in infants as young as 2 months.⁹⁹ Infants metabolize the analgesic isomer of ketorolac as fast as or faster than adults.¹⁰⁰

Opioids are a mainstay of pain management. They can be given through a variety of routes (oral, intravenous, neuraxial, intramuscular, and subcutaneous) and are very effective at reducing intraoperative and postoperative pain. Unfortunately, several adverse effects are associated with opioids, including nausea, vomiting, pruritus, constipation, respiratory depression, and rarely, death. Minimizing exposure to opioids is an important aspect of pain control and is a benefit of multimodal analgesia. An effective pain regimen is to provide scheduled (around the clock) nonopioid analgesics like acetaminophen. This will reduce pain scores and opioid consumption and possibly side effects.^{101,102} See Table 5-5 for analgesic medications and dosing.

Codeine has received recent attention because of its relationship with respiratory depression and death in children after tonsillectomy. The FDA placed a second black box warning on codeine in February 2013.¹⁰³⁻¹⁰⁵ Codeine is metabolized to morphine by the hepatic enzyme system CYP2D6. Patients with duplication of CYP2D6 metabolize codeine to significantly more morphine. This higher plasma level of morphine has resulted in significant postoperative complications. Many hospital systems have eliminated codeine from postoperative analgesia protocol for patients undergoing tonsillectomy and other pediatric surgical procedures. Oxycodone is not a prodrug like codeine and does not require metabolism to be analgesic. It is metabolized by CYP3A4, which has significantly fewer polymorphisms. It may be a more consistent and safer alternative.¹⁰⁶

Table 5-5 Analgesic Medications

Medications	Route	Dose
Acetaminophen	PO	15 mg/kg every 6 hr
	Rectal	40 mg/kg load, then 20 mg every 6 hr
	IV	15 mg/kg every 6 hr
Ibuprofen	PO	10 mg/kg every 6 hr
Ketorolac (toradol)	IV	0.5 mg/kg every 6 hr
Morphine	IV	0.02-0.1 mg/kg every 2-4 hr (prn)
Hydromorphone (Dilaudid)	IV	0.005-0.01 mg/kg every 2-4 hr (prn)
Oxycodone	PO	0.1 mg/kg every 3-4 hr (prn)

Multimodal therapy involves combining two or more of these medications. The benefit of multimodal therapy is improved analgesia with reduced side effects.

IV, Intravenous; *PO*, per os; *prn*, as needed (pro re nata).

Regional Techniques

Regional anesthesia can provide insensibility to a wide field and has become a popular technique to combine regional anesthesia with general anesthesia for pediatric plastic surgery patients. It includes neuraxial blocks (caudal and epidural), peripheral nerve blocks, and administration through wound catheters. Analgesia/anesthesia is achieved by placing local anesthetics and/or opioids into the epidural, intrathecal, or perineural space. In infants and children, peripheral and neuraxial analgesia is quite common. Intrathecal or spinal anesthesia is less common and is usually reserved for infants less than 45 weeks' PCA undergoing unilateral inguinal herniorrhaphy. Epidural and peripheral nerve blocks have been described for adult patients having plastic surgery.^{107,108} The benefit appears to be improved analgesia, decreased duration of hospitalization, and increased tissue oxygenation.^{109,110} Epidural anesthesia increases blood flow because of a local anesthetic-induced sympathectomy and may increase graft or flap survival.^{110,111} However, the price for increased blood flow may be a decreased mean arterial pressure, and this may compromise microcirculation to the flap. This phenomenon is more likely to occur in hypovolemic patients.^{112,113}

The reported benefits of the previously mentioned neuraxial blockade have been described for adults, information for pediatric patients is limited. Resting sympathetic vascular tone is diminished in infants, with minimal sympathectomy in those younger than 6 months of age.¹¹⁴ Despite this, epidural analgesia remains a very effective technique for managing acute postoperative pain in pediatric plastic surgery patients. Epidural and caudal blocks are a very effective form of analgesia for procedures involving both lower extremities or the abdomen or chest (Table 5-6). Urinary retention, nausea, and vomiting appear to be more common with neuraxial catheters than with peripheral nerve block catheters.¹¹⁵ Peripheral nerve blocks provide analgesia to a more specific nerve distribution, do not require a Foley catheter, and result in less nausea and vomiting than neuraxial catheters. Patients requiring upper or lower extremity surgery or abdominal or thoracic surgery can have their pain controlled with a peripheral nerve block. Catheters can also be placed, which will allow continuous analgesia for days. There is growing evidence that peripheral nerve catheters can be used to safely provide analgesia in the hospital and at home for ambulatory patients.¹¹⁶ Maximum allowable doses of common local anesthetics are listed in Table 5-7.

The risks of regional anesthesia vary according to the site, but in essence they include failure of the technique, bleeding at the site (such as epidural hematoma), infection, damage to underly-

Table 5-6 Regional Analgesia

Procedure	Block
Bilateral lower extremity reconstruction	Epidural block
Lower extremity flap	Peripheral nerve block (femoral, sciatic, and popliteal)
Upper extremity reconstruction or flap	Peripheral nerve block (interscalene, supraclavicular, infraclavicular, and axillary)
Iliac crest bone grafting	Paravertebral block, wound catheter

Table 5-7 Local Anesthetic Maximum Doses

Medication	Route	Dose
Lidocaine	Subcutaneous, neuraxial, peripheral nerve block	4 mg/kg (maximum dose) 7 mg/kg (maximum dose with epinephrine)
Ropivacaine	Subcutaneous, neuraxial, peripheral nerve block	3 mg/kg (maximum dose)
Bupivacaine	Subcutaneous, neuraxial, peripheral nerve block	3 mg/kg (maximum dose)

ing neural and nonneural structures, and inadvertent intravascular injection of a potentially toxic dose of local anesthetic. In addition, some patients may have physical characteristics, such as spina bifida or infection at the site, that contraindicate certain regional techniques.

Regional blocks are typically placed in anesthetized children. Rarely, the regional block may function as the primary anesthetic with supplemental sedation. For any procedure that uses a regional technique as the primary anesthetic, conversion to general anesthesia or emergent airway management is possible.

SPECIFIC PEDIATRIC PLASTIC SURGERY CONSIDERATIONS

Patients With Craniofacial Anomalies

Craniofacial anomalies are congenital or acquired deformities of the cranial and/or facial skeleton. Although rare, they make up a considerably diverse group of defects. The goal of surgical intervention is to restore both form and function. The classification of craniofacial anomalies is very difficult because of their variability, rarity, degree of severity, and lack of understanding of the etiologic factors and pathogenesis. The Committee on Nomenclature and Classification of Craniofacial Anomalies of the American Cleft Palate-Craniofacial Association¹¹⁷ has proposed the following classification:

1. Clefts
2. Synostosis
3. Hypoplasia
4. Hyperplasia
5. Unclassified

Clefts

Cleft lip is one of the more commonly recognized examples of a craniofacial cleft. However, craniofacial clefts involve a defect of the underlying cranial and/or facial skeleton, as well as the soft tissue envelope, and can involve the entire face. Tessier¹¹⁸ classified these defects by their locations, describing 15 various cleft positions. In this classification, the orbit is the center of the defect from which clefts radiate outward like the spokes of a wheel. *Treacher Collins syndrome*, also known as *mandibulofacial dysostosis*, is an example of a bilateral 6, 7, and 8 craniofacial cleft. Thompson first described Treacher Collins syndrome in 1846, and Treacher Collins later elaborated on the syndrome. It is characterized by poorly developed supraorbital ridges, aplastic/hypoplastic zygomas, ear deformities, cleft palate (in a third of cases), and mandibular and midface hypoplasia. From birth, the adequacy of the airway is the primary concern. The degree of airway obstruction is related to the degree of maxillary and mandibular hypoplasia, choanal atresia, and glossoptosis. Tracheostomy may be required during infancy for those at highest risk of OSA and sudden infant death syndrome.¹¹⁹ Aside from cleft lip and palate repair, major reconstruction typically is timed to occur during childhood or adolescence when the cranioorbitozygomatic bony development is nearly complete.

Anesthetic concerns specific to this syndrome primarily involve the airway. Infants and children with Treacher Collins syndrome may be very difficult or impossible to mask ventilate and/or intubate, and this airway difficulty can increase with age.¹²⁰ Several techniques have been successfully used to safely manage the airway in these infants. The LMA has been used to successfully ventilate a newborn infant with Treacher Collins syndrome for an extended period of time.¹²¹ Direct laryngoscopy, regardless of the blade used, may be difficult. However, the Bullard laryngoscope has been used to successfully intubate a child with Treacher Collins syndrome.¹²² The LMA and the air-Q LMA have also been used successfully to assist intubation of these children.¹²³⁻¹²⁵ Given the potential for difficult mask ventilation and intubation, this population may be best served with a sedated fiberoptic intubation. Another concern for anesthesiologists is protection of the patient's eyes. Because of the presence of maxillary and zygomatic hypoplasia, prone positioning may increase the risk of orbital compression and perioperative blindness. Infants and children with Treacher Collins syndrome can have congenital cardiac defects and require subacute bacterial endocarditis prophylaxis when indicated. Those most affected by upper airway obstruction and OSA may have reduced intraoperative and postoperative opioid requirements.³⁴

Craniosynostosis

Craniosynostosis is defined as an abnormal closure of one or more of the cranial sutures, creating abnormalities in the size and shape of the calvarium, cranial base, and orbits that constitute a diverse group of deformities. Craniosynostosis can also affect brain growth, ICP, and vision, resulting in developmental delay, increased ICP, and visual loss.

Craniosynostosis can occur by itself (simple) or as a major component of a syndrome (complex or syndromic). Six syndromes are associated with craniosynostosis: Apert syndrome, Pfeiffer syndrome, Saethre-Chotzen syndrome, Carpenter syndrome, Crouzon disease (also called *Crouzon syndrome*), and Muenke syndrome. Table 5-8 lists the various syndromes and their associated anomalies and anesthetic concerns. Infants and children with synostosis undergo surgery for cranial vault remodeling to reduce ICP, prevent brain injury, and enhance appearance. Repair of syndromic craniosynostosis may be more complicated and appears to be associated with increased blood loss. The specific cause of the increased bleeding is not known, but it might be related to the length of the operation.¹²⁶

The characteristic features of Apert syndrome include turribrachycephaly (high, steep, flat forehead and occiput), midface hypoplasia, and orbital hypertelorism. Cleft palate occurs in approximately 30% of cases. Choanal atresia and occasionally tracheal stenosis have been reported

Table 5-8 Anesthetic Considerations for Syndromic Craniosynostosis

Syndrome	Sutures	Clinical Features	Anesthetic Issues
Apert	Coronal	HEENT: Turribrachycephaly; midface hypoplasia; orbital hypertelorism; cleft palate in 30%; occasional choanal atresia and tracheal stenosis; airway obstruction Cardiac: Congenital heart disease occurs in 10%; may include VSD, pulmonary stenosis Genitourinary: Hydronephrosis in 3%; cryptorchidism in 4.5% Musculoskeletal: Syndactyly of hands and feet; fusion of digits 2-4; fusion of cervical vertebrae can occur Neurologic: Mental retardation common; may have elevated ICP Dermatologic: Acne vulgaris common	Preoperative laboratory values: Hematocrit, type and screen Airway management: Mask ventilation may be very difficult because of midface hypoplasia, choanal atresia, and tracheal stenosis; intubation may be difficult because of facial anomalies and decreased neck mobility Cardiac: Emphasis on balancing pulmonary and systemic blood flow; “de-air” IV lines; endocarditis prophylaxis Musculoskeletal: Cervical fusion may decrease neck extension; syndactyly may make vascular access difficult Neurologic: Caution with premedication if ICP is elevated
Pfeiffer	Coronal and occasionally sagittal	HEENT: Tower skull; midface hypoplasia; orbital hypertelorism; proptosis; choanal atresia is uncommon Pulmonary: OSA Cardiac: May have cardiac defects Musculoskeletal: Usually mild syndactyly involving broad thumbs and great toes; ankylosis of the elbow occurs rarely; fusion of cervical vertebrae reported Neurologic: Generally normal but mild developmental delay can occur; may have increased ICP	Preoperative laboratory values: Hematocrit, type and screen Airway management: No reported cases of difficult intubation; airway obstruction can occur intraoperatively or postoperatively Cardiac: Emphasis on balancing pulmonary and systemic blood flow; de-air IV lines; endocarditis prophylaxis Musculoskeletal: Cervical fusion may decrease neck extension; syndactyly may make vascular access difficult Neurologic: Caution with premedication if elevated ICP; eyes require protection if ocular proptosis present
Saethre-Chotzen	Coronal and others	HEENT: Brachycephaly; midface hypoplasia; orbital hypertelorism; beaked nose; occasional cleft palate Cardiac: May have cardiac defects Genitourinary: Renal anomalies and cryptorchidism Musculoskeletal: Short stature; mild syndactyly; cervical fusion possible Neurologic: Mild developmental delay; increased ICP is rare	Preoperative laboratory values: As in previous entries Airway management: No reported cases of difficulty with ventilation or intubation; however, patients may be at risk because of the airway and cervical pathology Musculoskeletal: Cervical fusion may decrease neck extension; syndactyly may make vascular access difficult Neurologic: Premedicate cautiously if ICP is elevated
Carpenter	Coronal and others	HEENT: Tower skull; downthrust eyes; orbital hypertelorism; low set ears; small mandible Cardiac: Cardiac defects common (VSD, ASD) Genitourinary: Hypogonadism Musculoskeletal: Syndactyly of hands and feet Neurologic: Developmental delay common but variable; may have increased ICP Other: Obesity	Preoperative laboratory values: As in previous entries Airway management: Small mandible may make intubation difficult; obesity may make ventilation difficult Musculoskeletal: Syndactyly may make IV access difficult Neurologic: Premedicate cautiously if ICP is elevated

ASD, Atrial septal defect; HEENT, head, ears, eyes, nose, throat; ICP, intracranial pressure; OSA, obstructive sleep apnea; VSD, ventricular septal defect.

Continued

Table 5-8 Anesthetic Considerations for Syndromic Craniosynostosis—cont'd

Syndrome	Sutures	Clinical Features	Anesthetic Issues
Crouzon	Coronal, lambdoid, and others	HEENT: Frontal bossing; tower skull; midface hypoplasia; beaked nose; hypertelorism; ocular proptosis; airway obstruction can occur Neurologic: Occasional mild developmental delay; may have increased ICP	Preoperative laboratory values: As in previous entries Airway management: Intubation may be difficult; may have airway obstruction during awake or sleep states; premedicate cautiously Neurologic: Premedicate cautiously if ICP is elevated; eyes require protection if ocular proptosis is present

HEENT, head, ears, eyes, nose, throat; ICP, intracranial pressure.

and can cause airway obstruction. Congenital cardiac disease is one of the more common associated visceral anomalies, occurring in approximately 10% of cases. Genitourinary anomalies (such as hydronephrosis and cryptorchidism) also occur in 10% of patients with Apert syndrome.¹²⁷ Severe craniosynostosis can result in increased ICP and, if uncorrected, developmental delay. Syndactyly of the hands and feet is often present, with fusion of digits 2 to 4, which can make intravenous access difficult. Cervical spine fusion has been reported¹²⁸; however, many children with Apert syndrome have been intubated successfully. In some cases suboptimal laryngoscopic views secondary to abnormal anatomy may require flexible fiberoptic intubation. The LMA may also be a reasonable adjunct for patients in whom intubation or ventilation of the lungs proves difficult. The air-Q LMA has been used as a conduit for intubation in infants and children with Apert syndrome.¹²⁹ The clinical features and anesthetic implications of Apert syndrome and the other acrocephalosyndactyly syndromes are presented in Table 5-8.

Unlike Apert syndrome, the other acrocephalosyndactyly syndromes are not typically associated with difficult airways. However, midface hypoplasia is common in these infants and may cause significant upper airway obstruction intraoperatively and postoperatively.¹³⁰ Crouzon disease, also called *craniofacial dysostosis*, is part of syndromic craniosynostosis. Infants with Crouzon disease have craniofacial anomalies without visceral or extremity involvement. The anomalies can result in significant airway obstruction that may require early tracheostomy. Table 5-8 outlines the main clinical features and anesthetic issues. During infancy, these patients undergo tracheostomy and/or cranial vault remodeling.

Hypoplasia

Hypoplasia of the craniofacial skeleton is a category of craniofacial anomalies characterized by hypoplasia or atrophy of a portion of the craniofacial soft tissue and skeleton. Pierre Robin sequence and hemifacial microsomia (including Goldenhar syndrome) are examples of these anomalies.

Pierre Robin sequence is characterized by retrognathia, glossoptosis (tongue falling to the back of the throat), and airway obstruction and probably occurs as a result of a fixed fetal position in utero that inhibits mandibular growth. Airway management of infants with Pierre Robin sequence can be very challenging because of difficulty with mask ventilation and intubation. Laryngeal masks have been successfully used to ventilate and assist in intubating these patients.^{57,58} Nasal intubation with a flexible fiberoptic scope has also been described.^{131,132} Direct laryngoscopy performed with a paraglossal approach can be very helpful in patients with small mandibles. This technique was described in infants with Pierre Robin sequence and was successful in five of the

six patients. The technique is described in the Airway Management section of this chapter (see Fig. 5-2). In infants who have significant difficulty with ventilation or intubation, aside from oropharyngeal and nasal airways, an 0 silk suture can be placed at the base of the tongue to displace it anteriorly for assistance with ventilation or intubation.

Hemifacial microsomia is characterized by unilateral or asymmetrical development of the facial bones and muscles and frequently involves the ear. It manifests as hypoplasia of the malar-maxillary-mandibular region and usually involves the temporomandibular joint. The vertebral pathology can involve the cervical vertebrae and can significantly reduce the cervical range of motion. Other associated anomalies of hemifacial microsomia include cardiac defects (such as ventricular septal defect, tetralogy of Fallot, and coarctation), renal defects, and neurologic defects (such as hydrocephalus). Patients with hemifacial microsomia can have significant upper airway obstruction and OSA.

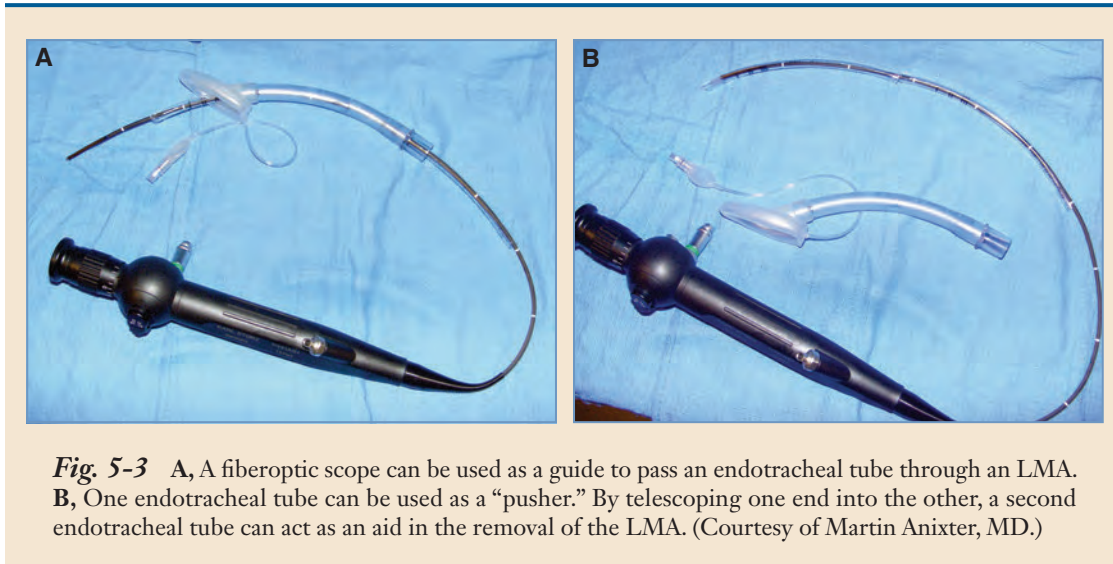
Airway management is a major concern in these patients. Mask ventilation may be difficult because of the facial asymmetry. Intubation is challenging because of the micrognathia and asymmetrical mandibular hypoplasia and potentially because of the decreased cervical range of motion. This difficulty may decrease with age but may increase after surgical reconstruction. Successful ventilation and intubation of infants has been reported using an LMA and fiberoptic laryngoscopy.¹³³

Anesthetic Management

The anesthetic management of infants with craniofacial anomalies begins with a complete preoperative evaluation. The history should define the anomaly and identify any associated syndromes. Infants and children with syndromes may have more complex airways, involvement of other organs, and more complicated surgical repair with more bleeding. Associated anomalies that can present a challenge to anesthesiologists include facial and airway features that make mask ventilation and intubation difficult. Airway pathology can also cause obstruction, and some of these children have OSA. A history of fatigue or sweating with feedings, cyanosis, and syncope are suggestive of an underlying cardiac anomaly. Cardiac abnormalities are associated with some syndromes, such as Treacher Collins syndrome, Apert syndrome, Pfeiffer syndrome, Carpenter syndrome, and hemifacial microsomia. In addition, some of these infants and children may have increased ICP that can manifest as headaches, vomiting, and somnolence, although chronically elevated ICP is often asymptomatic.

A thorough examination of the airway can be difficult in infants. Features that may predict difficulty with mask ventilation include midface hypoplasia and an enlarged tongue. In addition, a small mandibular space, decreased jaw opening and translocation, and decreased neck flexion and extension are predictive of difficult intubation. Additional examination should focus on identifying heart murmurs, and in infants with syndactyly, potential intravenous and arterial access sites. For reconstructions that involve significant blood loss, a preoperative hematocrit level should be obtained, and typing and crossmatch should be performed. Glucose levels should be monitored in formerly premature infants and infants younger than 2 months of age. Most children older than 1 year of age can be premedicated, but this is rarely necessary in those younger than 10 months old. Children with evidence of airway obstruction or increased ICP should be premedicated cautiously. Endocarditis prophylaxis should be considered for patients with congenital heart disease, per the American Heart Association guidelines.³⁸

Airway management in these patients can be very challenging. The difficulty may become evident during attempts at ventilation, intubation, or both. Fortunately, difficult airways are not common. However, the incidence is higher in patients with syndromes and in patients who have had previous reconstruction. Many techniques have been successfully described for infants and



children, and these include using a Bullard laryngoscope, LMA, air-Q LMA, flexible fiberoptic scope, and video laryngoscope.* A combination of techniques may be required to secure the airway. For example, an LMA has been used to facilitate passage of fiberoptic scopes and endotracheal tubes¹²⁴ (Fig. 5-3, A). An endotracheal tube/LMA size chart can be helpful for choosing the appropriate equipment to ensure success if an air-Q is not available. An air-Q LMA does not require an alteration of the endotracheal tube size. Both the tube and the pilot balloon can fit through the lumen of an air-Q LMA because of the removable cuff¹³⁵ (Table 5-9). The endotracheal tube can be secured while removing the LMA by telescoping a smaller endotracheal tube through the top of the larger endotracheal tube (Fig. 5-3, B). Commercial-grade “pushers” are available and are provided with the air-Q LMA. Cuffed endotracheal tubes may be more challenging to use with standard LMAs, because the balloon on the endotracheal tube may not fit through the lumen of the smaller LMAs. In this situation, uncuffed endotracheal tubes should be passed through an LMA of size 3 or smaller.

Preparation for Pediatric Procedures

Some infants with craniofacial anomalies require a tracheostomy because of significant upper airway obstruction.^{130,136} Adequate preparation involves having all of the necessary equipment available, with personnel trained and experienced in the use of these airway instruments. A pediatric otorhinolaryngologist may need to be immediately available.¹³²

Intraoperative Considerations

There are several intraoperative considerations for managing anesthetic for craniofacial repairs. Often these procedures are lengthy and expose infants to the risks of hypovolemia, hypothermia,

*References 58, 59, 65, 122, 129, 134.

Table 5-9 Laryngeal Mask Airway–Endotracheal Tube Compatibility

	LMA Size	Endotracheal Tube Size
Standard LMA	1.5	4.0 uncuffed
	2.0	4.5–5.0 uncuffed
	2.5	5.5 uncuffed
	3.0	6.0 uncuffed
	4.0	6.0 cuffed

	Weight (kg)	air-Q Size	Maximum Endotracheal Size (cuffed)
air-Q LMA	<7	1	4.5
	7–17	1.5	5.0
	17–30	2	5.5
	30–50	2.5	6.5
	50–70	3.5	7.5
	70–100	4.5	8.0

LMA, Laryngeal mask airway.

blood loss, and venous air emboli. Craniofacial procedures performed during an infant's first year of life include cranial vault remodeling, frontoorbital advancement, strip craniectomy (open and endoscopic), spring-mediated vault expansion, and distraction osteogenesis. Cranial-based procedures can involve significant blood loss because of the duration of the procedure and because of complications such as entering the sagittal sinus. Some studies suggest that 90% to 100% of these pediatric patients require blood transfusion.^{126,137} Endoscopic strip craniectomy, which is typically performed to correct sagittal synostosis, usually results in less blood loss.¹³⁸ Infants in particular are at risk for needing transfusions, because preoperatively they can be at the nadir of physiologic anemia (2 to 3 months of age), and the head contributes significantly more to the body surface area than in an older child or adult. Preparation for these procedures requires a baseline hematocrit level, blood type, and crossmatch. Adequate intravenous access must be obtained for resuscitation. In infants, at least two large-bore (18- and 22-gauge) peripheral intravenous catheters should provide adequate access. Arterial pressure monitoring is recommended for a beat-to-beat analysis of blood pressure and intravascular volume status and for blood gas monitoring. Some centers require central venous access for resuscitation and monitoring of central venous pressure.

Proposed techniques to minimize blood loss include surgical techniques and medical or pharmacologic techniques. Surgical techniques include scalp clips and needle-tip cautery. Medical techniques include preoperative autologous donation, acute normovolemic hemodilution, cell salvage, hypotension, antifibrinolytic therapy, and preoperative administration of recombinant erythropoietin (Epogen and Procrit). Most centers that perform cranial vault reconstructions did not use blood conservation techniques as of 2011.¹³⁹

Preoperative autologous donation and acute normovolemic hemodilution are techniques that have been described in older patients. No data currently support these techniques in infants

undergoing craniofacial surgery. The technique is limited in this population because of infants' small size, lower hemoglobin level, and reduced physiologic response to hemodilution, compared with that of adolescents.

Hypotensive anesthesia has been recommended for craniofacial surgery for decades, but it has not been well studied. A recent prospective study comparing two mean arterial blood pressures did not find a difference in the total amount of autologous blood given or the number of patients who were transfusion free.¹⁴⁰

In the past, the use of a Cell Saver system has been reported to be impractical for small pediatric patients because of the size of the collection reservoir. However, cell salvage reservoirs are now available in sizes as small as 25 ml. This technology may reduce the rate of autologous blood transfusions in infants undergoing craniofacial surgery. In a prospective observational study evaluating the use of a Cell Saver device with a 55 ml pediatric bowl for patients pretreated with erythropoietin, only 30% of the infants undergoing cranial vault remodeling required allogeneic blood, compared with 57% of historical controls.¹⁴¹

Antifibrinolytics have evolved significantly over the past decade. Aprotinin is no longer available because of the renal and myocardial complications associated with it in adults. Tranexamic acid and aminocaproic acid are alternatives that have filled the void. Two prospective blinded studies have demonstrated the efficacy of tranexamic acid. This antifibrinolytic reduces the volume of allogeneic blood exposure and increases the number of patients who are transfusion free.^{142,143} Although efficacy has been demonstrated, the safety of tranexamic acid is still being determined. Tranexamic acid has been associated with increased seizures after cardiac surgery¹⁴⁴; this seems to be dose dependent. In addition, this side effect has not yet been described after craniofacial surgery. The optimal dose of tranexamic acid has not been defined, but pharmacokinetic studies suggest that loading doses as low as 10 mg/kg with maintenance of 5 mg/kg/hr may be adequate to maintain a therapeutic plasma level.¹⁴⁵ The dose used in one of the efficacy trials was a 15 mg/kg loading dose, followed by 10 mg/kg/hr.

Preoperatively, recombinant erythropoietin decreases transfusion requirements for infants undergoing craniosynostosis repair. The reported dose of erythropoietin is 300 to 600 units/kg given subcutaneously once per week, along with oral iron supplementation. Erythropoietin is started 3 to 4 weeks before surgery. A prospective study of once-weekly dosing decreased the incidence of transfusion from 93% to 57% in infants undergoing craniosynostosis repair.¹⁴⁶

Prevention of Complications

Venous air embolism (VAE) is a potential complication of craniofacial and neurosurgical procedures. It can present with hemodynamic instability and result in death. VAE occurs commonly in pediatric patients having cranial-based procedures. A prospective study using precordial Doppler imaging in infants and children undergoing craniosynostosis repair detected VAE in 82% of patients. Hypotension secondary to VAE developed in 31%, but none developed cardiovascular collapse.¹⁴⁷ The incidence of VAE in this study was higher than the previously reported incidence of 66%.¹⁴⁸

Infants may be at increased risk for VAE, because significant hemorrhaging can occur during cranial vault remodeling, resulting in low central venous pressures. In addition, the relatively large size of an infant's head may raise the surgical site above the level of the heart, thereby increasing the pressure gradient for air entrainment. Some advocate placing central venous lines to monitor the trend of central venous pressures and minimize the risk of air embolism. However, to date there are no data to suggest that central venous pressure monitoring decreases the risk of VAE. Management of VAE begins with preventing hypovolemic states by providing adequate volume resuscitation and using precordial Doppler imaging for early detection of VAE. Lowering the



Fig. 5-4 Prone positioning for cranial vault remodeling. Plastic drapes from a Bair Hugger are applied. They act as a barrier to blood and irrigation. (Courtesy of Joseph Losee, MD.)

head of the bed, flooding the surgical field with saline solution, applying bone wax, discontinuing nitrous oxide, and providing inotropic support have been used for acute management of VAE.

Craniofacial procedures can be very lengthy, often lasting several hours. Complications resulting from long surgical procedures include skin breakdown, neuropathic injury, and hypothermia. Attention must be paid to the initial setup to ensure adequate positioning and padding to minimize these intraoperative injuries. Infants undergoing cranial vault remodeling may be placed in the prone position, with protection of the face and eyes.

Patients with syndromes that alter the architecture of the midface can present challenges when they are placed in the prone position, because it may be more difficult to adequately protect the face and eyes. An example of the initial setup is shown in Fig. 5-4. The infant is placed on a full-access forced-air warming system to minimize hypothermia, and the surgical site (the head) is isolated from the body with plastic drapes. This not only minimizes convective and radiant heat loss but also prevents conductive heat loss to a wet bed from irrigation and blood.

Postoperative Care

The postoperative management of infants undergoing craniofacial surgery depends on coexisting morbidities and the procedure performed. In patients who have had distractors placed, airway management may be more difficult after extubation because of the location of the device. Mask ventilation can be very difficult with external mandibular distractors in place. Airway equipment, including appropriately sized LMAs, should be available after extubation. External maxillary (midface) distractors are not typically placed in infants; however, their use in older children can make access to the airway more challenging; the presence of personnel and equipment in the operating room to remove part of the device is essential.¹⁴⁹

Infants having cranial vault remodeling and frontal orbital advancement can lose a significant amount of blood intraoperatively. If these patients are adequately resuscitated and hemodynamically stable, they can often be extubated in the operating room. Infants with difficult airways or significant airway obstruction and those who have had intraoperative complications may benefit from delayed extubation in the intensive care unit or operating room after their condition has stabilized.

Ongoing blood loss is common after major craniofacial surgery, and infants may require repeat transfusions in the immediate postoperative setting. Other complications that can occur

after craniofacial surgery include cerebral edema,¹⁵⁰ visual changes,¹⁵¹ cerebrospinal fluid leaks,¹⁵² infection,¹⁵² metabolic acidosis, and transfusion reactions.

Hyponatremia has been reported after major intracranial procedures and is common¹⁵³ but usually mild. It used to be attributed to syndrome of inappropriate diuretic hormone (SIADH). However, cerebral salt-wasting syndrome (CSW) may be the more likely cause. Both SIADH and CSW present with hyponatremia and high urine sodium levels. CSW is differentiated from SIADH by hypovolemia, high urine output, and low or normal antidiuretic hormone levels.¹⁴⁹ Proper diagnosis is important, because the management is different for the two syndromes. SIADH is treated with free water restriction, whereas CSW is managed with isotonic fluid replacement (normal saline solution) for maintenance and urine losses. Closely following the post-operative sodium levels will help guide therapy.

Patients With Epidermolysis Bullosa

Epidermolysis bullosa is a group of disorders of the stratified squamous epithelium that result in blistering, mucosal injury, and scarring after relatively minor mechanical forces.¹⁵⁴ The clinical effects range from minor disability with limited impact on functional capacity to death in the most severe cases. There are three major subtypes of epidermolysis bullosa: dystrophic epidermolysis bullosa, epidermolysis bullosa simplex, and junctional epidermolysis bullosa. They are defined by the dermal level in which blistering occurs. Patients with epidermolysis bullosa may undergo a variety of procedures that require the care of an anesthesiologist. Providing safe and effective anesthesia care for these patients can present significant challenges for the perioperative team.

The underlying pathology of epidermolysis bullosa affects many organ systems. Patients can develop blistering lesions of the mouth and oropharynx. The pain associated with these lesions can limit oral intake and result in nutritional deficiencies. Rarely, this results in dilated cardiomyopathy from selenium or carnitine deficiency.¹⁵⁵ A patient's poor nutritional status may also be reflected in abnormal electrolyte levels, compounded by ongoing fluid losses from blisters and denuded skin. Recurrent oral infections cause dental caries, oral scarring, and contractures that can limit mouth opening and may make intubation difficult. Scarring and contractures around the neck can also make intubation difficult by limiting neck mobility.¹⁵⁴

Recurrent injury to the esophagus leads to esophageal strictures, dysmotility, and gastroesophageal reflux. Recurrent episodes of streptococcal skin infections can lead to postinfectious glomerulonephritis with resulting renal dysfunction. The eyes can also be involved, and conjunctival bullae can develop with minimal mechanical stress. Anemia can occur in these patients.¹⁵⁶ The mechanism is unknown, but iron deficiency, poor nutritional intake, recurrent infections, and renal dysfunction may play a role. Treatment for epidermolysis bullosa may consist of topical and, rarely, oral steroids. Possible adverse effects of steroids include osteoporosis and adrenal suppression. Acute pain is common in infants and children with epidermolysis bullosa. Chronic pain ultimately develops in older patients, and this can result in chronic use of opioids.

Anesthetic Management

Common procedures performed for patients with epidermolysis bullosa include dental rehabilitation, esophageal dilation, Nissen fundoplication, skin biopsy and excision, and correction of pseudosyndactyly and contractures. Anesthetic management begins with the preoperative evaluation. The goal of the preoperative visit is to identify the severity of the disease and the nature of any coexisting diseases. Specifically, for patients with severe nutritional deficiencies, an echocardiogram and metabolic profile should be obtained to detect any underlying cardiac disease and/or electrolyte abnormalities. The degree and severity of gastroesophageal reflux needs to be

identified so that preparations can be made to minimize the risk of aspiration. For patients with a history of glomerulonephritis, blood urea nitrogen and creatinine levels should be checked to assess the degree of renal dysfunction. Anemia is common, and a preoperative hematocrit level should be obtained in most patients. Blood typing and crossmatching can be reserved for those who might require an intraoperative transfusion. Children who are chronically exposed to topical corticosteroids can have adrenal suppression and may require replacement steroids (2 mg/kg of hydrocortisone). Exogenous steroids given to replace the normal increase in cortisol requirements are referred to as *stress-dose steroids*.

Physical examination should identify patients with potentially difficult airways. For example, those with significant mouth contractures that limit mouth opening may be very difficult to intubate with laryngoscopy. The examination should also identify areas of skin that are particularly vulnerable or that have recently been injured. These should be documented and particular attention paid to positioning and protecting these areas.

Premedication can be beneficial in children with epidermolysis bullosa. Midazolam given either orally or through a gastric tube is very effective for reducing preoperative anxiety and may reduce the risk of injury to the skin during induction of anesthesia by facilitating patient cooperation. Intramuscular injections should be avoided. If an intravenous catheter is available, an H₂ receptor blocker (ranitidine [Zantac], 1 mg/kg) and metoclopramide (0.1 to 0.2 mg/kg) can be given to patients with gastroesophageal reflux disease. Adolescents with gastroesophageal reflux disease can also be given a nonparticulate antacid (Bicitra, 15 ml) orally or by gastric tube.

Transfer and Monitoring

Every effort should be made to minimize the patient's exposure to shear forces during table-to-table transfers. Lifting the patient on a flat, crease-free sheet and padding the surface of the bed and the patient's ankles with egg crate foam padding will help to protect against injury to the skin. Patients should never be transferred or repositioned by sliding. This shear force can cause significant injury.

Monitors can pose a challenge for patients with epidermolysis bullosa, because most monitors have adhesive coatings. ECG leads can be placed on the skin on the back after their adhesive coating has been removed. A cut-out square from a defibrillator pad can provide a nonadhesive barrier and will conduct an electrical signal.¹⁵⁷ Lubricated gauze such as Vaseline gauze can be placed on top of the leads to keep them in place. A pulse oximeter should also be secured over a finger after the adhesive backing has been removed. A clip pulse oximeter probe may be ideal and can be placed on the finger or earlobe. Likewise, temperature probes that are well lubricated can be placed in the esophagus. Noninvasive blood pressure cuffs may pose more of a challenge. For minor, short procedures, some suggest not using them. An arterial catheter can be used in place of a sphygmomanometer. Others have described the safe use of noninvasive blood pressure cuffs when they are placed on extremities that have been well padded with wool or polyvinyl chloride padding.^{154,157}

Induction

Induction of anesthesia can be achieved using an inhalation or intravenous technique. An intravenous catheter can be safely placed, but a tourniquet should not be used. Gentle circumferential pressure using a hand covered with a lubricated glove provides adequate venous engorgement. EMLA cream has been successfully used in children with epidermolysis bullosa.¹⁵⁸ A catheter can be secured with a nonadhesive dressing such as Vaseline gauze and a soft padded wrap (Fig. 5-5). The mask should be covered with a lubricant such as petroleum jelly or lubricated gauze (paraffin gauze). Induction should be calm and free from struggle. All airway equipment



Fig. 5-5 Intravenous placement and protection are facilitated by the application of a soft cotton wrap. A pulse oximeter is applied without the use of adhesive backing. (Courtesy of Larry Borland, MD. Permission granted by family for photography.)



Fig. 5-6 Nasal intubation in a patient with epidermolysis bullosa. The tube is secured with lubricated gel foam. (Courtesy of Larry Borland, MD. Permission granted by family for photography.)

(laryngoscope, oral and nasal airways, and endotracheal tubes) should be well lubricated. If possible, the endotracheal tube should be one-half size smaller than typically would be used.

A fiberoptic scope may be necessary to successfully intubate some patients with epidermolysis bullosa. Generally, oral intubation is recommended, but nasal intubation can be safely performed with well-lubricated and appropriately sized nasal tubes (Fig. 5-6). The endotracheal tube can be secured with an external “arm” that is attached to the bed or with lubricated gauze that is gently wrapped around the patient’s head. Oral and pharyngeal lesions have occurred after intubation, but the incidence is not high if appropriate measures are taken.¹⁵⁹ The use of an LMA has been described in patients with epidermolysis bullosa. Ames et al¹⁵⁷ reported the use of the LMA on 57 occasions, with only one case of a new lingual bulla.

Postoperative Care

Postoperative pain management for patients with epidermolysis bullosa may be difficult because of the combination of acute and chronic pain. Opioids can be given intravenously in the form of

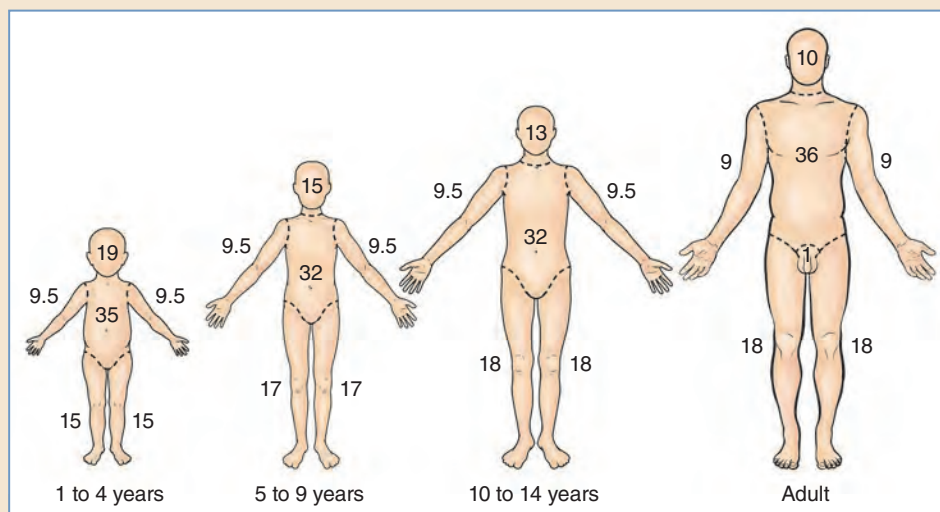


Fig. 5-7 Proportion of TBSA in relation to age (Rule of Nines). The contribution of the head to the body surface area is larger in infants and children.

patient-controlled analgesia. Regional analgesia has been successful for postoperative pain control, and peripheral nerve blocks such as brachial plexus, axillary, and ankle blocks have also been described.¹⁶⁰⁻¹⁶² Placing peripheral nerve blocks is complicated by the poor integrity of the skin.

The skin preparation should be sprayed or poured on without any wiping. No adhesive drapes can be applied, and some investigators have advocated not using nerve stimulators, because the positive electrode is applied with an adhesive patch. Alternatively, a percutaneous needle may be used. The catheter can be secured to the skin with sutures. Neuraxial blocks with caudal, epidural, and intrathecal anesthesia have also been described.^{163,164}

Patients With Burn Injury

Burns occur in approximately 1.3 million children every year, and 3000 children die each year from them, making burns the second leading cause of traumatic death in childhood.¹⁶⁵ The most common cause of burns in this age group is hot liquid scalds. Other causes include flame or thermal heat, electricity, and chemical and radiation exposure.

The depth and the percentage of body surface area involved define the severity of the burn. The components of body surface area in pediatric patients are different from those in adults. The head contributes more to the body surface area in infants and children, and the extremities contribute less¹⁶⁶ (Fig. 5-7).

The injuries from burns can affect almost every organ system. The skin is the primary barrier against dehydration, fluid and electrolyte loss, and bacterial infection. Burns destroy this barrier and expose the body to these risks. The initial insult to the skin results in an inflammatory response and causes tissue edema. Cytokines, arachidonic acid metabolites, and oxygen free radicals mediate the edema. Fluid loss from the intravascular space to the surrounding tissue can be significant, and edema may develop at sites remote from the burn. This edema fluid is rich in albumin and in a moderate to large burn can account for profound albumin loss over the course of a few days.¹⁶⁷

The initial cardiovascular response to a burn is decreased cardiac output. The decrease occurs because of a reduction in preload (intravascular volume loss) and an increase in systemic vascular resistance and PVR.¹⁶⁸ Direct myocardial depression results from circulating factors such as tumor necrosis factor, vasopressin, and oxygen free radicals. The decrease in cardiac output is quickly replaced in hours to days by an increase in cardiac output (two to three times the baseline) after the burn. After the initial injury, a hypermetabolic state develops that lasts until the wounds are closed. This state is a neuroendocrine response fueled by an increase in catecholamines, glucocorticoids, glucagon, and cytokines. Aside from an increase in cardiac output, hypertension is common.

Lung injury from burns is a significant cause of morbidity and mortality and can occur in the upper or lower airway. Heat exposure to the larynx can cause edema and obstruction. Increased capillary permeability can occur at sites remote from the burn and may contribute to upper and lower airway edema. Any evidence of a burn to the head or neck or evidence of soot on the face should raise suspicion of an airway injury. Interventions to secure the airway should not be delayed, especially in patients with coughing, wheezing, or stridor. This is more critical in pediatric patients, because small changes in the radius of their airways will cause a significantly greater increase in airway resistance.¹¹ (Resistance is inversely proportional to the fourth power of the airway radius.)

Inhalation injuries can result from exposure to carbon monoxide and hydrogen cyanide. Smoke inhalation during fires, especially in closed spaces, increases this risk. Carbon monoxide avidly binds hemoglobin (200 times greater than oxygen) and decreases the ability of hemoglobin to deliver oxygen to the tissues, resulting in asphyxiation. The half-life of carbon monoxide is reduced from 4 hours to 40 minutes by exposing the patient to high concentrations of oxygen.^{165,169} Carboxyhemoglobin levels should be measured in all patients being treated for burns that occurred in a house fire or a fire in a closed space. These patients should be treated with high-concentration ($\text{FiO}_2 = 1$) oxygen. Cyanide poisoning should be suspected with all inhalation injuries. Treatment consists of sodium thiosulfate and sodium nitrite, although care must be taken with sodium nitrite, because it can cause hypotension.^{170,171}

Anemia of thermal injury is common after burns. Its cause is multifactorial and includes hemorrhage, hemolysis, and decreased erythroid production.^{172,173} The initial loss of red cells occurs as a result of bleeding and hemolysis. The surviving erythrocytes have a decreased half-life that appears to be related to a circulating inhibitor, which explains the reduced half-life of transfused blood as well. Erythrocyte production is also decreased. Erythropoiesis is diminished because of an inhibitor of erythroid stem cells, which can result in profound anemia.¹⁷²

The physiologic changes that occur after burns affect the pharmacokinetics and pharmacodynamics of many drugs. Pharmacokinetic changes include alterations in distribution and clearance. The tissue edema that occurs after burns increases total body water and increases the volume of distribution. Larger doses of intravenous medications may be required to achieve adequate plasma levels. A hypermetabolic state results in hepatic enzyme induction and may enhance drug clearance. Protein binding is altered as well. Albumin levels decrease significantly after burns because of intravascular losses to surrounding tissue. Conversely, alpha-1-acid glycoprotein is an acute-phase reactant and increases after burns. Drugs that bind to these proteins (for example, acidic and neutral drugs, which bind with albumin, and basic drugs such as bupivacaine, fentanyl, and alfentanil, which bind with alpha-1-acid glycoprotein) have altered free drug plasma levels.

Clinical Management

Thermal injuries in adults and children result in a decreased sensitivity to nondepolarizing neuromuscular blockers and an increased sensitivity to depolarizing neuromuscular blockers.¹⁷⁴ Larger doses of nondepolarizing neuromuscular blockers are required to achieve adequate clinical results

because of the increased extrajunctional acetylcholine receptors.^{175,176} The duration of neuromuscular blockade with nondepolarizing blockers is also diminished. Larger doses of nondepolarizing neuromuscular blockers shorten the onset time and prolong the duration of neuromuscular blockade but do not achieve the onset time and duration seen in nonburned patients.¹⁷⁷ Depolarizing neuromuscular blockers such as succinylcholine (Anectine and Quelicin) should not be used, because the increased number of acetylcholine receptors may cause hyperkalemia. It is not known how soon after a burn this sensitivity begins, but succinylcholine should probably not be used 24 hours after a burn has occurred. This effect can persist for years after a burn.

Surgical Management

Surgical management of burn patients depends on the depth of the burn. Superficial burns can be managed with dressing changes. Partial-thickness burns may require skin grafting if they do not heal after 2 to 3 weeks of dressing changes. Full-thickness burns require skin grafting. Surgical debridement of burned tissue can result in significant bleeding. There are several descriptions of extensive or massive hemorrhage during surgical debridement.¹⁷⁸ Pediatric patients have smaller blood volumes and a larger surface area/weight ratio and are at particular risk of significant blood loss. Many methods have attempted to minimize blood loss, including subcutaneous and topical vasoconstrictors, tourniquets, normovolemic hemodilution, and induced hypotension.¹⁷⁹ Despite these techniques, significant bleeding can persist. A study using a dermabrader for surgical debridement of partial-thickness burns and needlepoint electrocautery for full-thickness burns resulted in significantly less bleeding. Some patients did not require transfusions.¹⁸⁰ In this study the donor sites were also infiltrated with saline solution and epinephrine. Doses of epinephrine of up to 10 µg/kg were used without adverse effects such as arrhythmias. In pediatric burn patients, doses greater than 10 µg/kg of subcutaneous epinephrine have been well tolerated.^{181,182}

Anesthetic Management

Anesthetic management of all burn patients begins with the airway. Evidence of inhalation injuries should prompt early intubation before airway edema occurs. Cuffed endotracheal tubes can be used safely in pediatric patients of all ages and should be considered in these patients, because high peak airway pressures may be required to successfully ventilate them. All patients with inhalation injuries need to be evaluated for carbon monoxide and hydrogen cyanide poisoning and should be treated with high-concentration oxygen.

Immediately after a burn, a patient can develop profound hypovolemia caused by hemorrhage and intravascular volume loss to surrounding tissue. The Parkland formula and Brooke formula help to guide the initial fluid resuscitation for the first 24 hours¹⁸³ (Box 5-1).

Box 5-1 Modified Parkland and Brooke Formulas for Pediatric Burn Reconstruction

Parkland Formula

$[4 \text{ ml/kg crystalloid} \times \% \text{ TBSA burned} \times \text{Weight (kg)}]^* + \text{Maintenance fluids}^\dagger$

Brooke Formula (modified)

$[0.5 \text{ ml/kg crystalloid} + 1.5 \text{ ml/kg colloid} \times \% \text{ TBSA burned} \times \text{Weight (kg)}]^* + \text{Maintenance fluids}^\dagger$

*Administer the first half of the volume over 8 hours and the second half over 16 hours.

†Maintenance fluids = 4 ml/kg/hr for the first 10 kg + 2 ml/kg/hr for the second 10 kg + 1 ml/kg/hr for every kg over 20 kg. TBSA, Total burned surface area.

However, these estimates are not completely accurate for managing infants and children. Children have a higher surface area/weight ratio. Formulas that base resuscitation on percentage of surface area burned and weight may underestimate the fluid requirements in children. In children younger than 4 years of age, calculating the fluid requirements from the previously mentioned formulas and then adding their daily maintenance requirements has been suggested.¹⁸⁴ Fluid losses in burned pediatric patients may be profound, exceeding 4000 ml/m² of burned skin, compared with 2500 ml/m² in adults.^{183,185}

During surgical debridement of full-thickness and partial-thickness burns, patients can have massive blood loss and fluid shifts. Surgical techniques, as described earlier in the chapter, can minimize bleeding. Regardless, the need for transfusions is very common, and adequate vascular access and monitoring are essential before surgery. This may include placing arterial and central venous catheters. Frequent blood gas analysis should be performed to assess the adequacy of resuscitation and ventilation. Many burned patients can have low ionized calcium levels that persist for weeks.¹⁸⁶ Ionized calcium should be measured and replaced, especially during resuscitation with citrated blood products.

Burn patients become poikilothermic because of the significant evaporative heat loss that occurs from open wounds. This is especially true in pediatric patients, because their body surface area is relatively larger. Every effort must be made to minimize heat loss when these patients come to the operating room. Steps to decrease heat loss include raising the operating room temperature as high as tolerated by the operating room staff, radiant warming lights, forced warm air devices, and giving warm intravenous fluids. Wrapping the extremities and head in plastic may significantly decrease heat loss.

Adequate pain management is critical for pediatric burn patients. They have pain not only from the burn but also from therapeutic maneuvers such as repositioning, dressing changes, and surgical procedures. Opioids are the mainstay of pain management for burns. Most opioids have been prescribed for managing burns, including morphine, methadone (Dolophine and Methadose), and remifentanyl (Ultiva); however, morphine is the most common. The pharmacokinetics of opioids in burn patients is inconsistent, and few data are available for children. In three adult studies, the pharmacokinetics of morphine did not differ significantly from that of nonburned patients.^{187,188} However, Furman et al¹⁸⁹ found the volume of distribution and clearance to be smaller in burned patients, and the elimination of morphine was prolonged, compared with elimination in nonburned adults. Opioids can be delivered as a continuous infusion or through patient-controlled analgesia. Children older than 5 years of age start to understand the concept of patient-controlled analgesia. By 7 years of age, most children can use patient-controlled analgesia effectively. Other modalities have been used for breakthrough pain caused by dressing changes and surgical debridement. Ketamine is a very effective analgesic, and its safety profile is excellent. The hallucinations associated with its use can be very disturbing for children, but these can be reduced by giving a benzodiazepine such as midazolam. Ketamine may have the added advantage of reducing opioid tolerance and opioid hyperalgesia.

Regional anesthesia has been described for children with burns. Both neuraxial and peripheral nerve blockade have been reported and can provide very effective analgesia.^{190,191} The potential advantages are excellent analgesia and reduced exposure to systemic opioids. The disadvantage is that the location and extent of the burn may preclude placing a regional catheter. For appropriate cases, this technique should be considered.

KEY POINTS

General

- General anesthesia for infants and children is very safe.
- The concerns regarding the neurocognitive effects of anesthesia/sedation on the developing brain are not clearly defined.
- Recent prospective evidence did not identify any negative neurocognitive effect of general anesthesia on the developing brain.

Pediatric Physiology

- A neonate's heart (and to some degree an infant's heart) has a limited ability to increase stroke volume; neonates are poorly tolerant of bradycardia. Moreover, significant volume loads may more easily cause ventricular overload and failure.
- Pediatric patients are more prone to hypoxia because of (1) the larger closing capacity (the sum of the closing volume and the residual volume; *closing volume* is the volume above the residual volume at which airway closure/collapse occurs in dependent lung zones), (2) more chest wall compliance, and (3) higher oxygen consumption.
- Neonates and infants are at increased risk of thermoregulatory instability, because they are at a higher risk of heat loss, and they have a decreased ability to produce heat.

Preoperative Evaluation

- Many craniofacial anomalies warrant concerns regarding difficult airways, OSA, and congenital heart disease, and these patients should be seen preoperatively by the anesthesiology team.
- Infants with a PCA of less than 45 to 50 weeks are at risk for apnea after general anesthesia and need to be observed overnight with an apnea/bradycardia monitor.
- Patients with OSA and sleep-disordered breathing can have disordered breathing and opioid sensitivity while recovering from the sedative effects of anesthesia. They may be poor candidates for same-day surgery.
- Children who have upper respiratory infections with fevers above 38° C (100.4° F), changes in behavior, purulent nasal drainage or cough, or wheezing during lung auscultation may benefit from having their surgeries postponed for 2 to 6 weeks.

Continued

KEY POINTS (continued)

Preoperative Evaluation—cont'd

- Separation from the family and introduction of a face mask can cause tremendous anxiety in children and may contribute to postoperative behavioral changes such as bed-wetting, increased separation anxiety, and nightmares. Anxiety can be reduced by giving midazolam 0.5 to 1 mg/kg, which should be used with caution in patients with severe upper airway obstruction or increased ICP.

Anesthetic Management

- Induction of anesthesia in a pediatric patient can be achieved using an inhalation or intravenous technique.
- Equipment that can be used to secure the airway in anesthetized children includes but is not limited to a mask, LMA (air-Q), an endotracheal tube, a laryngoscope, a flexible fiberoptic scope, and a video laryngoscope.
- Sedation from recent anesthetic and pain medications given in the recovery room can cause hypoventilation and airway obstruction. Airway obstruction from swelling of the airway and the soft tissues surrounding it may occur either in the operating room or in the recovery room.
- PONV is the most common immediate cause of postoperative anesthetic morbidity in children.
- Children may develop emergence delirium postoperatively, characterized by confusion, inconsolability, psychomotor excitement, and dysphoria.
- Epidural and peripheral nerve blocks have been described, and the benefit may be improved analgesia, decreased duration of hospitalization, and increased tissue oxygenation.

Craniofacial Surgery

- Craniofacial anomalies are classified as follows: (1) clefts, (2) synostosis, (3) hypoplasia, (4) hyperplasia, and (5) unclassified.
- Treacher Collins syndrome is an example of a craniofacial cleft and may present with a difficult or impossible airway, upper airway obstruction, and congenital heart disease.
- Anesthetic concerns during craniofacial surgery include potentially difficult airways (ventilation and intubation), significant blood loss, venous air emboli, and hypothermia.
- Techniques described to minimize blood use include needlepoint electrocautery, scalp clips antifibrinolytic therapy, cell salvage, and synthetic erythropoietin.

- Postoperative concerns after craniofacial surgeries (cranial vault reconstructions) include ongoing blood loss, metabolic acidosis, transfusion reactions, and hyponatremia.

Epidermolysis Bullosa

- The underlying pathology of epidermolysis bullosa affects many organ systems and may cause cardiac dysfunction, renal failure, reflux, malnutrition, anemia, and chronic pain.
- Children who are chronically exposed to topical corticosteroids can have adrenal suppression and may require stress-dose steroids.
- Patients should not be transferred or repositioned by sliding. Lifting patients on a flat, crease-free sheet and padding the surface of the bed and ankles with egg crate foam padding will help to protect against injury to the skin.
- Monitors can pose a challenge for patients with epidermolysis bullosa. The following measures can help to prevent complications:
 - ECG leads can be placed on the skin or on the back after their adhesive coating has been removed.
 - The pulse oximeter should be secured over a finger after the adhesive backing has been removed.
 - Temperature probes that are well lubricated can be placed in the esophagus.
 - Blood pressure cuffs are safe when placed on extremities that have been well padded with wool or PVC padding.
- All surfaces that come in contact with the skin need to be well lubricated to minimize shear forces (including masks, gloves, laryngoscopes, LMAs, and endotracheal tubes).
- Airway management can be challenging in patients with epidermolysis bullosa because of limited mouth opening and neck mobility.

Burn Surgery

- The injury caused by burns can affect almost every organ system.
- Edema can develop at sites remote from the burn.
- The initial cardiovascular response to a burn is decreased cardiac output. This is quickly replaced by an increase in cardiac output (two to three times the baseline) hours to days after the burn and lasts until the wounds are closed.

Continued

KEY POINTS (continued)

Burn Surgery—cont'd

- Any evidence of a burn to the head or neck or evidence of soot on the face should raise suspicion of an underlying airway injury. Interventions to secure the airway should not be delayed, especially in patients with evidence of coughing, wheezing, or stridor.
- Inhalation injuries can result from exposure to carbon monoxide and hydrogen cyanide.
- Anemia of thermal injury is common after burns.
- The hypermetabolic state results in hepatic enzyme induction and may result in enhanced drug clearance. Protein binding is altered, because albumin decreases and alpha-1 acid glycoprotein increases after burns.
- Larger doses of nondepolarizing neuromuscular blockers are required to achieve adequate clinical results because of the increased extrajunctional acetylcholine receptors.
- Depolarizing neuromuscular blockers such as succinylcholine should not be used for 24 hours after a burn, because the increased number of acetylcholine receptors may cause hyperkalemia.
- Surgical debridement of burned tissue in children can cause significant bleeding leading to massive hemorrhage.
- A technique for surgical debridement using a dermabrader for partial-thickness burns and needlepoint electrocautery for full-thickness burns may result in significantly less bleeding.
- Burn patients are poikilothermic because of the significant evaporative heat loss from open wounds.
- Adequate pain management is critical for pediatric burn patients and may consist of opioids, ketamine, and regional anesthesia.

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6

Pediatric and Fetal Wound Healing

Jacqueline S. Israel • Timothy W. King



he process of normal cutaneous wound healing in infants, children, and adolescents is very similar to that of adults. It is a highly complex process, and when it is affected by various local and systemic factors, healing may go awry. In contrast to postnatal wound healing, fetal healing is a distinct, unique process that investigators are beginning to understand. This chapter outlines cutaneous wound healing in pediatric patients, offers examples of factors that contribute to abnormal healing, introduces what is currently known about fetal wound healing, and provides an initial approach to wound care in infants and children.

WOUNDS IN INFANTS AND CHILDREN

A wound results from damage to normal tissue. The mechanism of damage may vary; in the infant and pediatric populations, wounds are often acute. Plastic surgeons may encounter many types of wounds in an infant, child, or adolescent, as described in Table 6-1.¹⁻³

Once a wound has been created, *wound healing* is a complex series of events that lead to repair. *Cutaneous wound healing* consists of a coordinated, complex series of events that can be thought of as a “temporal and spatial” process.⁴ The temporal component begins at the time of injury and continues for months to years as the scar matures. Healing can also be conceptualized spatially, occurring simultaneously on molecular, cellular, tissue, organ, and system levels.⁴

Table 6-1 Examples of Wound Types

Wound	Description
Abrasion	Frequently involves only the epidermis; often heals by secondary intention
Bite	Should have a high index of suspicion for infection (for example, polymicrobial, rabies, tetanus)
Chemical burn	Can occur from povidone-iodine, alcohol-based washes, adhesive removers
Crush	Difficult to assess wound severity; monitor for signs of compartment syndrome
Extravasation	Common in neonates and infants; caused by infusions (for example, crystalloids, electrolytes, chemotherapy)
Laceration	Ideally closed within 8 hours, or up to 24 hours if the face is involved
Maceration or “moisture-associated skin damage”	As seen with diaper dermatitis; can lead to hypergranulation, infection ³
Pressure ulcer	Should have a high index of suspicion in hospitalized infants and children; frequent position changes are paramount
Puncture	Should have a high index of suspicion for a foreign body; often heals by secondary intention
Sting	May result in systemic effects requiring epinephrine, steroids, or antihistamines
Surgical incision or wound	Often a clean wound that can be closed primarily
Tear/blister	More common in pediatric patients (thin skin); often iatrogenic
Thermal burn	Can be caused by hot water, light therapy, metal objects (electrodes, household items), fire; infants and children have different body surface area proportions compared with adults

NORMAL CUTANEOUS WOUND HEALING IN INFANTS AND CHILDREN

One should have at least a basic knowledge of normal healing to understand and approach clinical wound healing challenges. Normal wound healing is characterized by four discrete but overlapping stages: coagulation, inflammation, proliferation, and maturation (Fig. 6-1).

Coagulation

The process of *coagulation* begins with tissue injury, and hemostasis is the primary goal (Fig. 6-2). Platelets aggregate and form a fibrin plug (clot) through alpha-degranulation and the release of adhesive proteins, such as fibrinogen, fibronectin, thrombospondin, and von Willebrand factor.^{5,6} This clot acts as an initial matrix on which wound healing will occur. There is activation of both the intrinsic and extrinsic clotting cascades, which facilitates hemostasis and converts fibrinogen into fibrin, turning the clot into a gel-like matrix to facilitate cell migration. Platelets also release platelet-derived growth factor (PDGF) and transforming growth factor-beta (TGF-beta), which chemoattract inflammatory cells such as neutrophils and monocytes. After hemostasis has been achieved, local blood vessels vasodilate, promoting the influx of inflammatory cells. Activation of the complement cascade (C3a and C5a anaphylatoxins) increases blood vessel permeability, which also recruits inflammatory cells to the wound bed.⁶

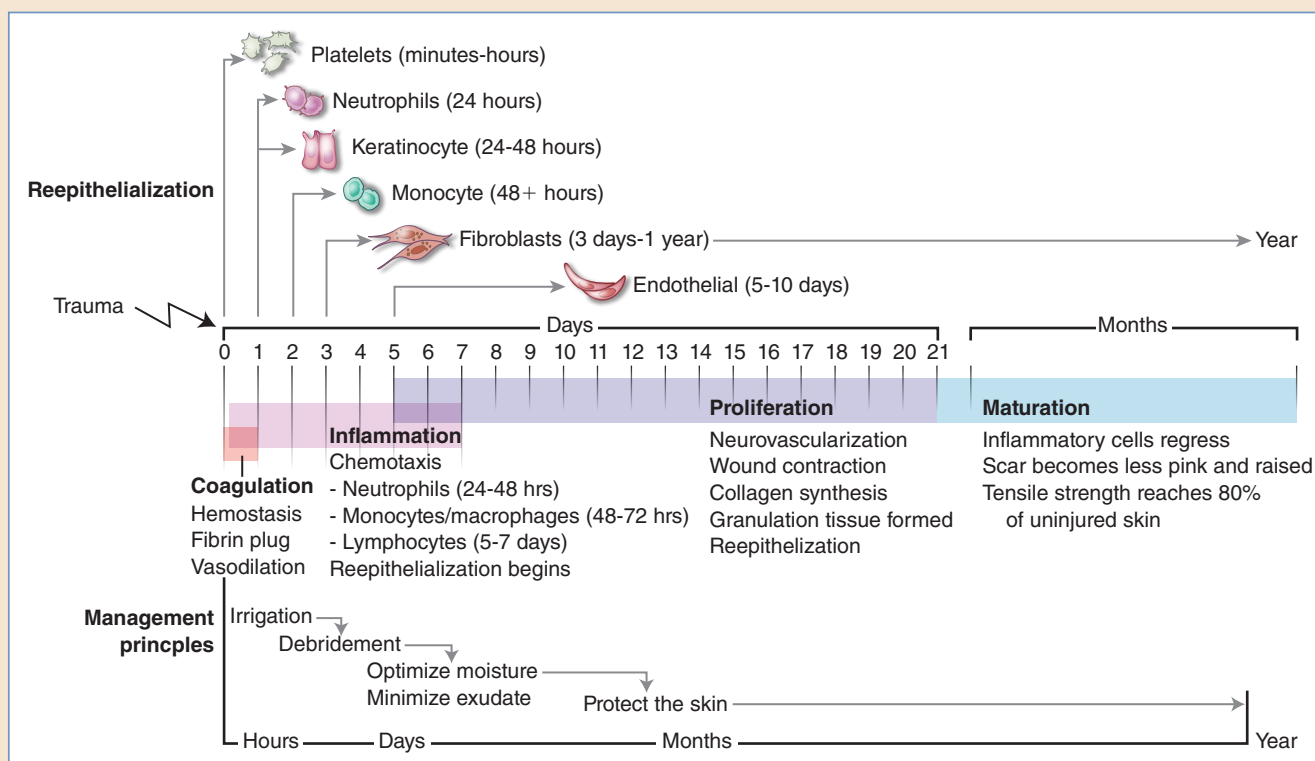


Fig. 6-1 Stages of normal cutaneous wound healing.

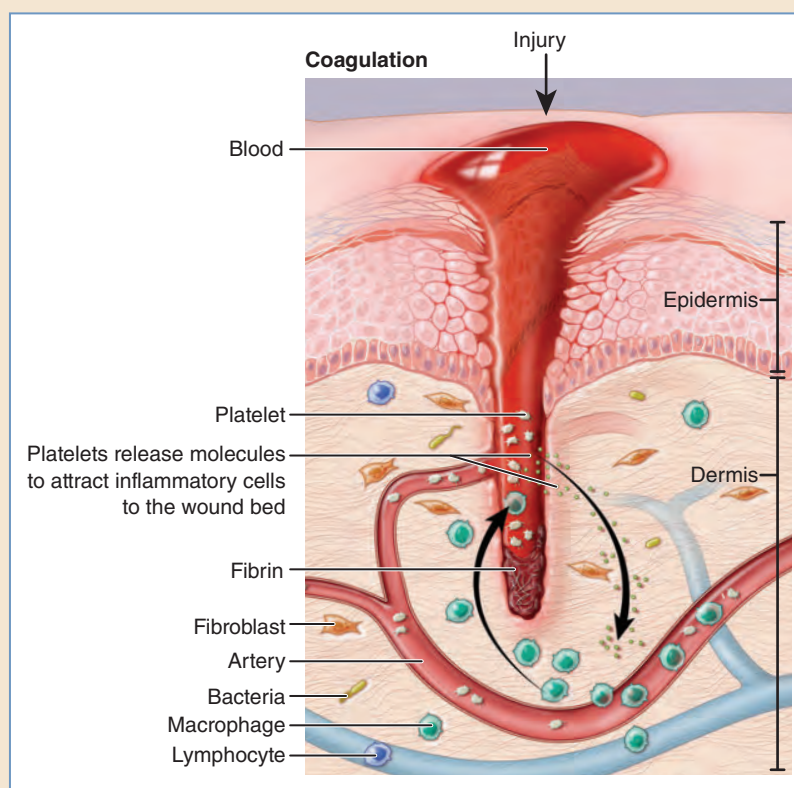


Fig. 6-2 The process of coagulation.

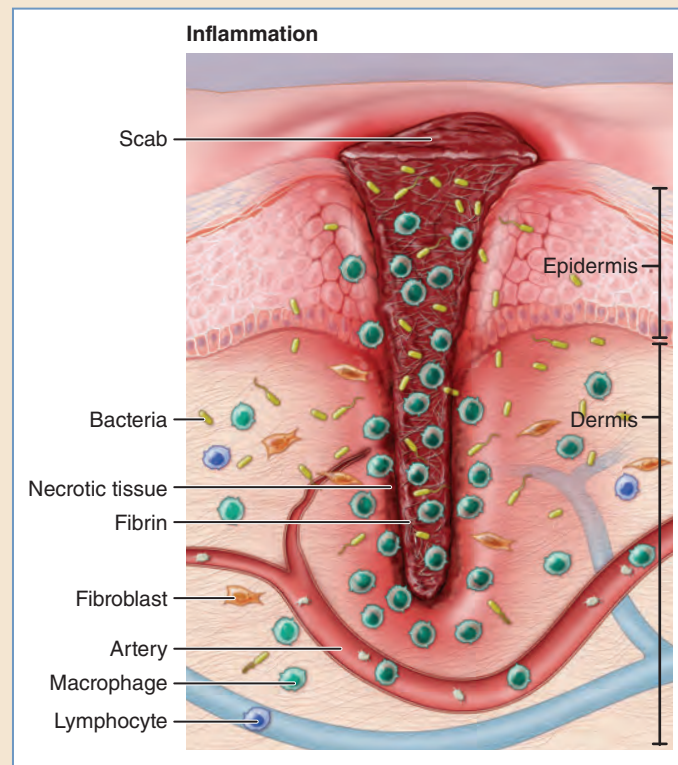


Fig. 6-3 Inflammation.

Inflammation

The next step in the wound healing process is *inflammation*, which is characterized by a flood of inflammatory cells into the wound. The primary goal of this process is to rid the wound of bacteria, debris, and devitalized tissue (Fig. 6-3). The first to enter, within 24 to 48 hours after injury, are the polymorphonuclear leukocytes, or neutrophils.⁶ Neutrophils are recruited to the wound by TGF-beta1, PDGF, tumor necrosis factor alpha, and interleukin-1. Neutrophils exhibit oxidative burst, an oxygen-dependent process that kills bacteria through free radicals. They also cause phagocytosis of debris and release collagenases to degrade and remodel the wound matrix. When the wound becomes clean, neutrophils exit the wound bed. This is important, because if bacteria and debris persist, the neutrophils remain and may cause damage to nearby healthy tissue.⁶ Recent studies suggest that neutrophils are not essential for wound healing.⁷ This is in contrast to the role of the next cell to enter the wound, the monocyte; wounds will not heal properly if they are not present.⁷

Monocytes are the predominant cell type in the wound by 48 to 72 hours after injury.^{5,6} They are recruited to the wound by monocyte chemoattractant protein-1. On arrival, monocytes become activated and transform into macrophages. Macrophages scavenge the wound for foreign material, bacteria, and debris and release additional growth factors that activate endothelial cells, fibroblasts, and keratinocytes. They also participate in tissue degradation by secreting collagenases (matrix metalloproteinase [MMP]-1, MMP-9, and MMP-3).⁸ Lymphocytes are the final cells to enter during the inflammatory stage, appearing around 5 to 7 days after tissue injury. Their role is less clear.

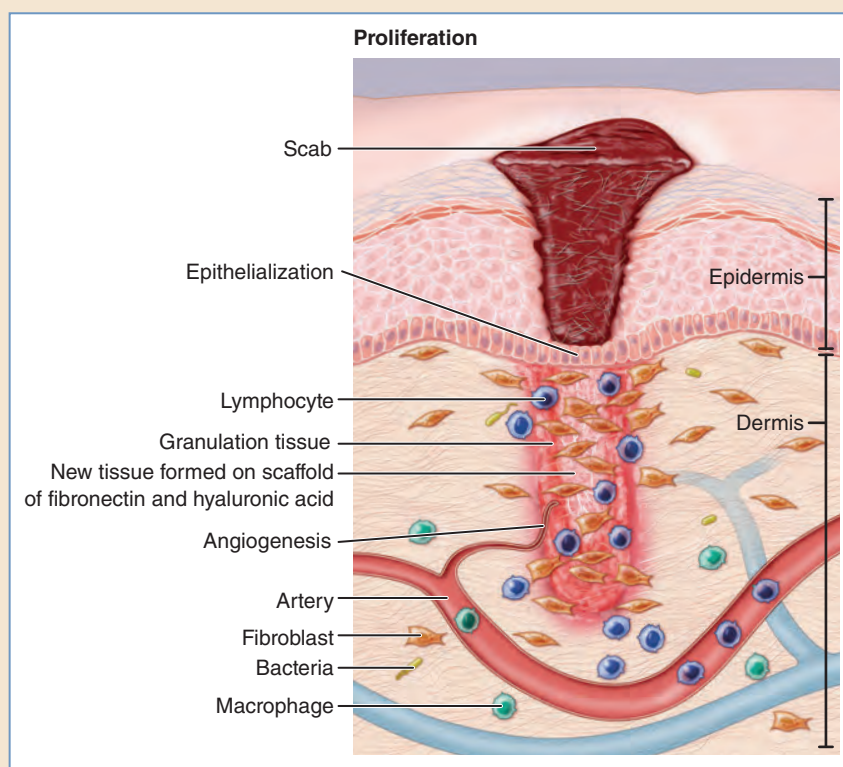


Fig. 6-4 Proliferation.

Proliferation/Matrix Deposition

As inflammation declines, the process of *proliferation* begins, typically around day 4, and continues for up to 3 weeks. It is named for the migration of fibroblasts and generation of new tissue at the wound bed. During this phase, there is angiogenesis, collagen deposition, formation of granulation tissue, epithelialization, and wound contraction (Fig. 6-4).

Fibroblasts predominate beginning about days 3 to 5.⁶ Infants and children have been found to have higher numbers of fibroblasts during this phase, perhaps explaining the faster rate of wound healing often observed in this population.² Initially, there is a period of *fibroplasia*, whereby fibroblasts are brought to the wound by PDGF and TGF-beta. After migration to the wound, they begin to secrete extracellular matrix (ECM) components, such as loosely organized collagen and hyaluronic acid (HA). The adhesion proteins fibronectin, laminin, and tenascin are essential for fibroblast migration and attachment.⁶ Eventually, the platelet-fibrin clot (established during the coagulation phase) is replaced by a scaffold of fibronectin and HA, which is the basis for new tissue formation.

Collagen synthesis occurs on fibroblast ribosomes. Chains of collagen are assembled, and hydroxylation of specific prolines and lysines occurs. Ascorbic acid (vitamin C) is required for the enzyme prolyl hydroxylase to convert proline to hydroxyproline. After the individual collagen fibrils are synthesized, the enzyme lysyl oxidase organizes collagen molecules into collagen fibrils through covalent cross-linking.⁶ Both type I and type III collagen are synthesized. Collagen has many isoforms with subtle differences in molecular structure. These differences are responsible for the vast differences in the structures and functions of particular collagen molecules. For

example, bone is primarily type I collagen, whereas skin is approximately 80% type I and 20% type III collagen. Normal blood vessels are 80% type III and 20% type I collagen.

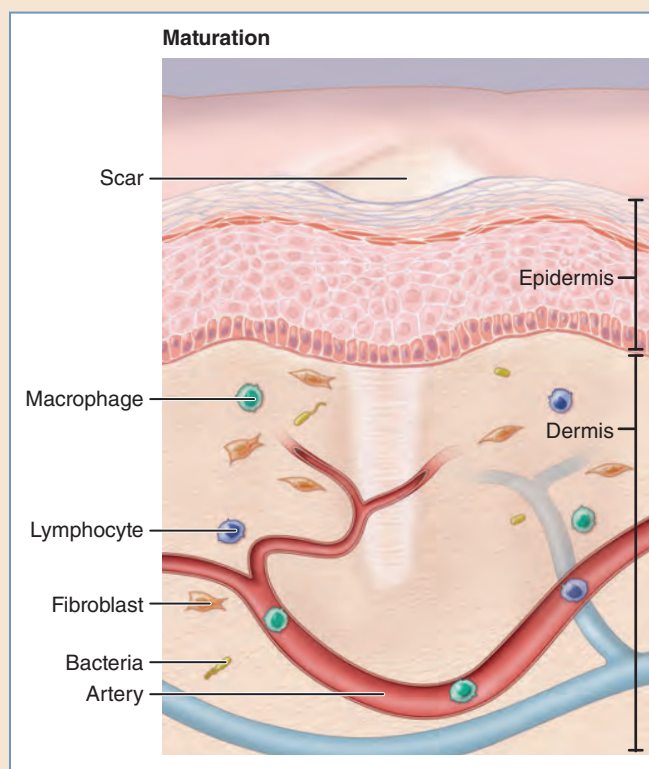
Another process during the proliferation phase is wound *contraction*. Open wounds decrease in size by contracting, minimizing the need for new tissue. Around week 2, some of the collagen-synthesizing fibroblasts transform into *myofibroblasts*, which facilitate narrowing of the wound by contracting along the edges of tension. *Neovascularization* also occurs. Fibroblast growth factor (FGF)-2 from macrophages stimulates endothelial cells to release tissue plasminogen activator and procollagenase, which digest the ECM and allow immature tubes of endothelial cells to grow into the wound. Vascular endothelial growth factor stimulates endothelial cell proliferation, and TGF-beta facilitates the differentiation of the vascular endothelial cell type. Alongside the process of angiogenesis, *granulation tissue* is formed. This process is primarily stimulated by PDGF and vascular endothelial growth factor. Granulation tissue is characterized by a dense bed of capillaries, fibroblasts, and immature ECM, and it seems to form more quickly in neonates and children than in adults.²

Reepithelialization of the wound begins as early as 24 to 48 hours after injury but is an important part of the proliferation phase.^{1,6} Keratinocytes at the wound edges become migrating keratinocytes and contain integrin receptors and actin filaments to facilitate migration and recreate a cellular barrier overlying the immature and evolving wound matrix (which contains collagen, fibrin, fibronectin, tenascin, and vitronectin).^{5,6,9} Both keratinocytes and fibroblasts secrete laminin and collagen IV, facilitating the progressive construction of a new basement membrane.⁶ Many growth factors and cytokines are involved in reepithelialization, including hepatocyte growth factor, FGF-7 and FGF-10, and transforming growth factor-alpha.⁵

Maturation

Wound remodeling, or *maturation*, occurs over weeks to months after injury. In this stage, inflammatory cells regress, and the scar becomes less pink and raised (Fig. 6-5). Tensile strength ultimately reaches approximately 70% to 80% that of normal, uninjured skin.⁹ By 3 to 5 weeks after the injury, fibroblasts are at the center of the wound and synthesize primarily type I and type III collagen. The process of collagen deposition and degradation (via MMP-1 and MMP-3) occurs concurrently for up to 1 year, and collagen becomes more densely packed as time progresses. FGF-1 and FGF-2 facilitate remodeling as the wound matures.

Compared with scars in adults, scars in children remain pink and elevated for a longer time. In addition, scars formed during the first 6 months of life become almost imperceptible, in contrast with scarring in older children. The reasons for this are not clear. This phenomenon may be related to two factors: (1) During the third trimester of fetal healing, scars become virtually absent, perhaps as a holdover from some of the phenomena from this period, and (2) in the transition from fetal to infant healing, there is a change in the predominance of collagen types (type III to type I). The unique aspects of fetal healing are discussed in more detail later in the chapter.

Fig. 6-5 Wound maturation.

ABNORMAL CUTANEOUS WOUND HEALING IN CHILDREN

Aberrant wound healing can result from both local and systemic factors.¹ In response to these factors, wound healing in any organ system reflects a balance between tissue regeneration and scar formation.⁹ In the case of pediatric cutaneous wound healing, scar formation predominates, and the formation of an organized scar is a normal biologic response to tissue injury.

As discussed by Gurtner and Wong,⁹ wound healing failure can be attributable to (1) inadequate regeneration of previously normal tissue (as seen with injuries to the central nervous system), (2) inadequate scar formation (as often seen in chronic wounds, such as those in patients with pressure ulcers and persistent inflammatory states), (3) excessive regeneration (as in the case of a neuroma), or (4) excessive scar formation (as in a hypertrophic scar or keloid). Any wound in a pediatric patient can be analyzed within the context of the local and systemic factors that affect the balance between regeneration and scar formation.

Issues Related to Aberrant Wound Healing

Fluid Collections and Edema

Edema can delay and preclude wound healing, particularly after the acute inflammatory response.² Excess fluid in and near a wound can present as a seroma or hematoma, which may result in ischemia or infection. Just as in adults, edema in children can be addressed with elevation, compression, and negative pressure wound therapy (NPWT) when appropriate.¹

Pressure

Mechanical pressure impairs normal wound healing mechanisms through local shearing forces and decreased blood flow. Compared with adults, infants and children have thin skin and are susceptible to pressure ulcers both in dependent areas and in tissue that is in contact with medical devices.³

Ischemia

Decreased oxygen delivery to tissues from ischemia is detrimental to wound healing.¹ Oxygen is required for aerobic metabolism, macrophage oxidative burst, fibroblast proliferation, and collagen production. In children, this is often most relevant in the setting of hypoperfusion related to a systemic process, such as sepsis, blood loss, dehydration, or hypothermia. Arterial and venous sufficiency can be readily assessed at the bedside by examining the degree of capillary refill, tissue warmth, distal pulses, and the presence of bleeding at the cut edges of the wound.

Bacterial Burden

Normal wound healing mechanisms become impaired when the bacterial count in a wound exceeds 10 colony-forming units per gram of tissue.^{1,5,10} Exacerbating factors include a retained foreign body, the presence of necrotic tissue, and poor hygiene. Thorough irrigation and debridement can help prevent infection; antibiotics should be administered when an infection is either expected or suspected.

Contracture

Wounds close through the mechanism of contraction, but contracture may occur when normal, elastic soft tissue is replaced by scar, resulting in an aesthetic and functional abnormality. Any wound in a pediatric patient may lead to contracture over time. Examples include linear wounds or incisions over joints (where the development of a contracture can impair mobility), burns, and Volkmann ischemic contractures (attributable to muscle ischemia).

Hypertrophic Scars and Keloids

Hypertrophic scars and keloids result when the normal wound healing processes of proliferation and remodeling are exaggerated both in time and space, with too much dermal collagen deposited. Both are known to occur with greater frequency in the pediatric age group, with the incidence peaking in the second decade.^{11,12} Excessive scarring in children may result in decreased quality of life and can negatively affect psychosocial well-being and self-confidence.⁴

Hypertrophic scars are red, raised, dermal lesions that can develop after any form of tissue injury.^{13,14} They do not grow or extend beyond the border of the original wound, and the lesion is often well demarcated with a linear outline. Hypertrophic scars typically appear 4 to 8 weeks after injury, display rapid growth for up to 6 months, and resolve with time.¹¹ Clinically, hypertrophic scars can cause pruritus, especially when located on high-tension areas, such as the shoulder, neck, presternum, knee, and ankle, but tend not to be painful.¹⁴ Often the best treatment is to reassure patients and parents that the lesions will resolve without intervention. *Keloids* (derived from the Greek word *cheloides*, meaning “crab’s claw”) are raised dermal lesions that extend beyond the boundaries of the original wound.^{11,13,14} Histologically, keloids are composed of disorganized type I and type III collagen. Keloids may present years after the initial injury and may even develop spontaneously, particularly in areas of tension or in those patients who experience friction from clothes or accessories. Keloids are firm, shiny, pinkish-purple in color and may have telangiectasias. They too are well demarcated, but the borders are often quite irregular. The most common locations include the anterior chest, shoulder, earlobe, arms, and cheeks. Although keloids appear more frequently in individuals with darkly pigmented skin, investigators have not yet discovered a clear genetic pattern or a specific gene related to keloids. Like hypertrophic scars, they cause pruritus, but may also cause significant pain, burning, and hypersensitivity.¹¹

In a 2009 review, Juckett and Hartman-Adams¹¹ offered grade B recommendations (limited-quality patient-oriented evidence) regarding the treatment of hypertrophic scars and keloids: (1) Cryotherapy can be considered for smaller keloids and in combination with other treatments; (2) intralesional corticosteroid injections are appropriate first-line modality for the treatment of both keloids and hypertrophic scars; (3) pressure dressings can be used for the prevention of hypertrophic scars, particularly in the setting of burns; and (4) combination therapy is a second-line approach when first-line modalities fail to offer an improvement (for example, excision, silicone sheeting, or steroid injection).

Perhaps the most popular treatment is intralesional injection of triamcinolone (10 to 40 mg/ml, depending on location).^{11,15} This can reduce the size of the lesion and relieve some of the burning and itching. It should be administered with caution in pediatric patients because of the risks of adrenal suppression, depigmentation, and skin atrophy. Silicone sheeting has been used with variable efficacy.^{12,16} Excision of keloids is often disappointing because of the high recurrence rate (as high as 75%). Nevertheless, surgical debulking or total excision may be required for functional or aesthetic reasons.

Protein-Calorie Malnutrition

Suboptimal nutritional status impairs wound healing in infants and children. One should establish a baseline nutritional status in any child presenting with a wound; this includes asking about weight loss, dietary intake, food intolerance, and known illnesses.¹ Children with wounds in the setting of acute illness require aggressive nutritional supplementation for appropriate healing.

Vitamin Deficiency

Vitamins A, B, C, and K, zinc, copper, iron, magnesium, omega fatty acids, and the amino acids arginine and glutamine are essential for proper wound healing. Deficiencies should be identified and addressed.¹

Genetic Disorders

Inherited abnormalities in tissue architecture can negatively impact wound healing. Individuals with connective tissue abnormalities may heal more slowly, be more susceptible to injury, or demonstrate aberrant healing mechanisms. Examples of such disorders include Ehlers-Danlos syndrome, Marfan syndrome, and osteogenesis imperfecta (OI).¹ Ehlers-Danlos syndrome represents a spectrum of genetic defects characterized by abnormal production or processing of collagen. Common clinical findings reflect a lack of normal collagen and include hypermobile joints, hyperelastic, thin, and fragile skin, and blood vessel abnormalities. Marfan syndrome is an autosomal dominant disorder that results from an abnormality in fibrillin, which is essential for ECM formation. Clinical manifestations include joint hypermobility, hyperelastic skin, and aortic aneurysms with the risk of rupture. Finally, OI is characterized by a deficiency of type I collagen, and bone is most affected. There are multiple types of OI; in all types, wound healing may be delayed or impaired. A suspected connective tissue disorder in a child necessitates prompt referral to a geneticist, and wound management should be initiated with a multidisciplinary, specialized approach.

Diabetes

Both type 1 and type 2 diabetes mellitus can occur in children. One of the detriments to wound healing in patients with diabetes is local tissue ischemia and poor perfusion of the skin and subcutaneous tissue. Chronic diabetic wounds likely have more acute inflammation but relatively poor epithelialization, granulation, collagen proliferation, angiogenesis, and overall wound strength.¹⁷ It has been suggested that elevated hemoglobin A1c is associated with delayed wound healing; thus optimal glucose control is essential.¹⁸

Immunodeficiency

Inflammation and the immune response are part of normal cutaneous wound healing. Patients with acquired or inherited immunodeficiency are at increased risk of poor wound healing. Some examples of acquired immunodeficiency include the use of corticosteroids, chemotherapy, and infections, such as HIV. Inherited immunodeficiency syndromes that may contribute to abnormal wound healing in pediatric patients include chronic granulomatous disease, common variable immunodeficiency, and severe combined immunodeficiency. Neonates and infants have a less mature immune system than that of adults, which may precipitate infection, skin damage, and difficulties with wound healing.²

FETAL WOUND HEALING

In 1979, while studying limb amputations presumably attributable to amniotic banding, Rowlatt¹⁹ observed that fetal cutaneous wounds can heal without forming scar tissue. This initial discovery was a catalyst for the scientific drive to characterize fetal wound healing at the molecular, cellular, tissue, and clinical levels.

Fetal skin undergoes a rapid period of development, beginning at week 3 of gestation, when the epidermis is first formed.²⁰ The epidermis becomes keratinized at week 14, is considered to have all components of mature adult skin at week 16 (for example, dermal appendages and basal layer), and throughout the remainder of the second and third trimesters, the skin undergoes further maturation to the point where it is felt to be identical to adult skin by birth²⁰ (Fig. 6-6). It is hypothesized that, up to at least week 26 of gestation, fetal wound healing is primarily regenerative (for example, scarless)⁷ (Fig. 6-7).

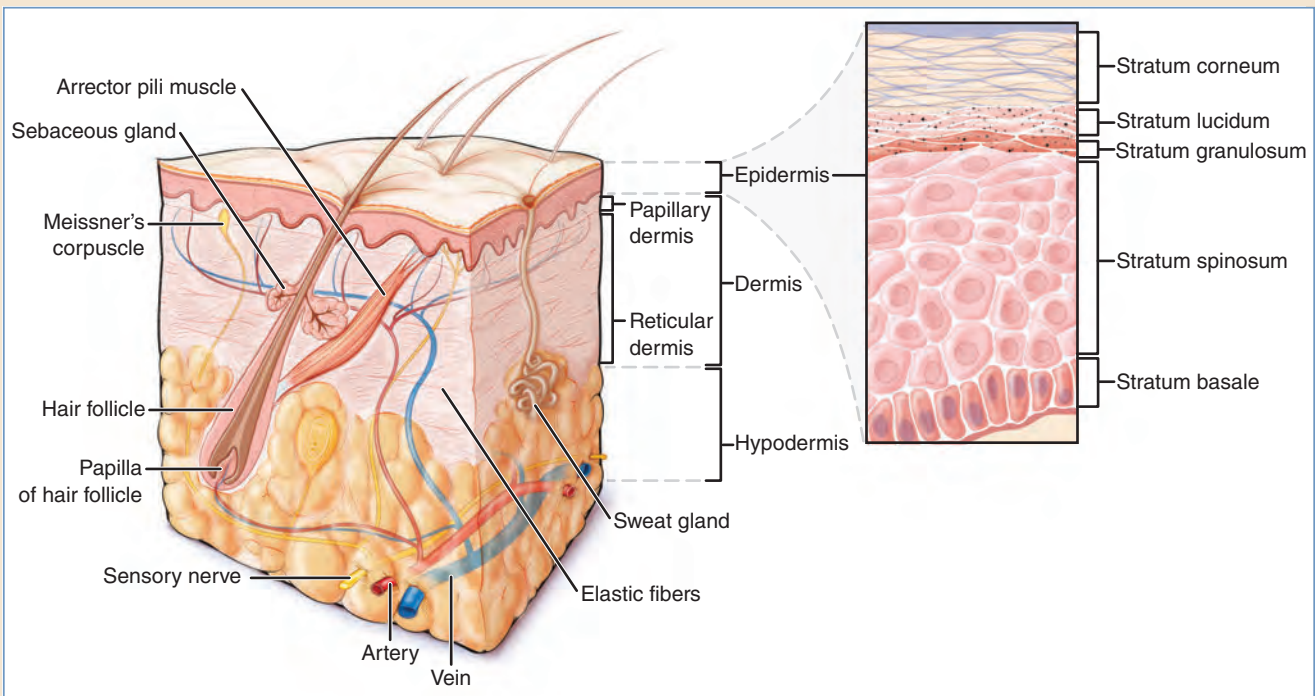


Fig. 6-6 Normal pediatric skin.

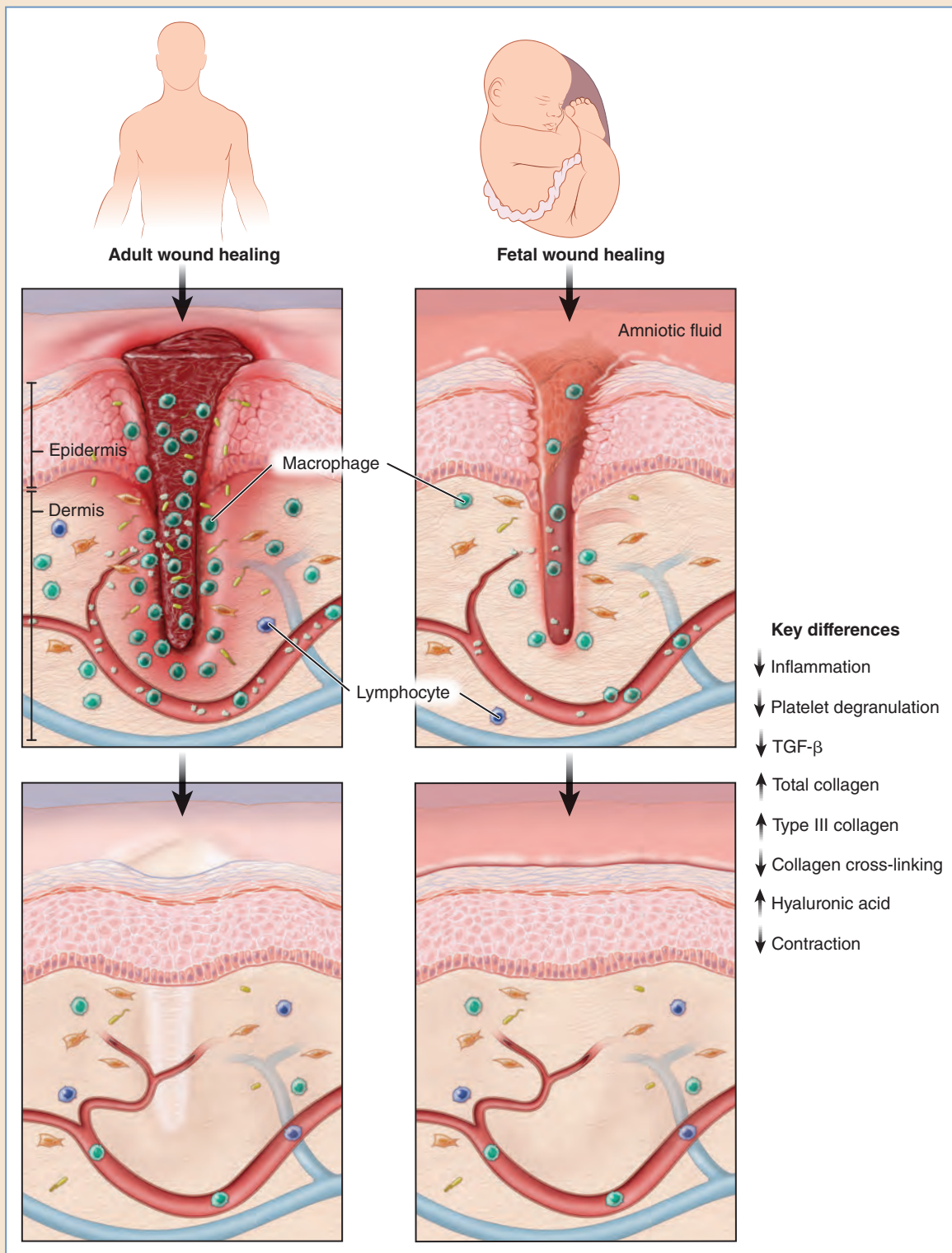


Fig. 6-7 Fetal wound healing compared to adult wound healing.

Differences in inflammatory cells and mediators, ECM (including collagen synthesis and deposition), and gene expression seem to account for many of the known differences observed in fetal and adult cutaneous wound healing.^{2,20-23} It was initially hypothesized that the ability of wounds to heal in utero without scarring was attributable to amniotic fluid, but we now know that there are intrinsic differences in fetal skin that allow it to heal without perceptible scars.^{21,24-27} Although there is a great deal of unknown regarding fetal healing, Table 6-2 lists the key characteristics of fetal wound healing that have been identified to date.

Table 6-2 Molecular, Cellular, and Tissue Differences in Fetal Healing Compared With That of Adults

Differences	Characteristics of Fetal Healing (compared with adult healing)
Inflammation ^{7,20,21,23,27}	
General response	↓ Inflammatory response ↓ Number of immune cells
Platelets	↓ Platelet degranulation
Cytokines	↑ IL-10 (blocks inflammation) ↓ IL-6 and IL-8
Neutrophils	↓ Adhesion molecule expression leads to ↓ neutrophil migration to wound bed ↓ Prostaglandins
Growth factors ^{7,20,21,28,29}	↓ VEGF (↓ inflammation and ↓ scarring) ↓ PDGF ↓ FGF ↓ TGF-β1/↓ TGF-β2 (↓ inflammation, ↓ scarring) ↑ TGF-β3 (↓ inflammation) ↓ IGF-1
ECM ^{7,20,21,23,30-33}	
Fibroblasts	Fibroblasts act differently (produce collagen and proliferate simultaneously)
Collagen	↑ Total collagen ↑ Type III and IV collagen ↑ Type III:type I collagen ↑ Prolyl hydroxylase ↓ Collagen cross-linking Fine, reticular pattern of collagen deposition (compared with dense, parallel bundles)
HA	↑ HA, HA receptors
Proteoglycans	↓ Decorin, ↓ heparin sulfate ↑ Chondroitin sulfate ↑ Fibromodulin (blocks collagen fibrillogenesis, blocks TGF-β)
Other	↑ MMPs (favor ECM turnover and remodeling)
Adhesion molecules ^{20,21,23,33}	↑ Fibronectin (attachment) ↑ Tenascin (migration)
Wound closure ^{21,22,28,34}	Myofibroblasts act quickly, briefly (if at all) ↓ Wound contraction (may be caused by amniotic fluid) ↑ Actin cables for closure
Cell-signaling transcription and gene expression ^{21,22}	Differences in phosphorylation ↑ Homeobox (Hox) gene expression regarding growth and proliferation

ECM, Extracellular matrix; FGF, fibroblast growth factor; HA, hyaluronic acid; IGF, insulin-like growth factor; IL, interleukin; MMPs, matrix metalloproteinases; PDGF, platelet-derived growth factor; TGF, transforming growth factor; VEGF, vascular endothelial growth factor.

Not all fetal wounds heal scarlessly. The degree of scar formation depends on several variables, including size (for example, larger wounds are more likely to heal with a scar), location (for example, skin versus intestinal tissue), species (scarless healing has been observed in fetal mice, rats, and humans; less so in fetal sheep and rabbits), and time (increased gestational age results in a lower likelihood of scarless healing).²⁰ Although there have been many important observations and discoveries regarding fetal healing on the molecular and cellular level, we have yet to acquire knowledge that significantly changes our clinical approach. The hope is that with time, the collective body of information acquired from fetal healing experiments will facilitate a clinical breakthrough in promoting scarless healing in both children and adults.

MANAGING WOUNDS IN INFANTS AND CHILDREN

Pretreatment Assessment

Wound prevention is an essential consideration in neonates and children, particularly in hospitalized patients. Because infant skin is thin and susceptible to tears and blistering, vigilant assessment for pressure injuries in the occipital region and from external objects, such as tracheostomies, gastrostomies, extremity and trunk braces, and wheelchairs, is paramount.³⁵ Frequent position changes (for example, every 2 hours) can help minimize friction and the risk of pressure ulcers.

Acute wounds are common in pediatric patients. As with any injury, the initial assessment prioritizes any life-threatening injuries and the ABCs (airway, breathing, circulation).¹ Hemostasis should be an initial goal with respect to soft tissue injury. A thorough history and physical examination in a pediatric patient should focus on the nature of the wound (acute versus chronic, the time course, mechanism of injury, degree of contamination, size, zone of injury, and presence of devitalized tissue). Other factors to be assessed include medical conditions, medications, previous injury or radiation, and social/family situation. Laboratory studies may be obtained if there is a suspicion of a systemic abnormality (for example, infection or malnutrition) or if there is hemodynamic instability. An initial workup for acute wounds often includes radiographs or other imaging (particularly for bites and penetrating, crush, degloving, and injection injuries), although one should be cognizant of the risks of radiation in children.

Wound Care Considerations

There are several mechanisms by which a wound in a child can heal. One way, as is often the case with surgical wounds, is primary intention (for example, primary closure). Delayed primary closure may be most useful when there is a great deal of tissue edema and in traumatic wounds in which there is questionable tissue viability.¹ Secondary intention involves allowing an open wound to close through granulation and reepithelialization. This can be performed with traditional dressing changes or by using NPWT. Skin grafts (either split-thickness or full-thickness) and/or flaps may be necessary when there is insufficient tissue coverage to facilitate adequate healing by primary or secondary intention.¹

Other considerations include the use of topical ointments and dressings, antibiotics, and tetanus and rabies prophylaxis.^{1,36-38} With respect to ointments and topical products, maintaining a moist healing environment is most important for optimizing granulation and epithelialization.³⁵ Most pediatric wounds do not necessitate antibiotic therapy if they are thoroughly irrigated and debrided. Exceptions include bites or penetrating wounds, grossly contaminated wounds, and perioperative antibiotic prophylaxis.³⁶ Tetanus prophylaxis should be administered to all children and adolescents whose vaccinations are not up to date or who have not yet received their full schedule of tetanus immunization.³⁷ Consideration should be given to the age of the child,

because specific formulations of tetanus vaccination may be indicated (for example, a booster containing tetanus toxoid as well as diphtheria toxoid and acellular pertussis)³⁷:

- Clean wounds: Tetanus toxoid is given if the child has received fewer than three previous doses or if the immunization history is unknown, or if it has been more than 10 years since the most recent booster.
- Grossly contaminated wounds and complex wounds: Tetanus toxoid and immune globulin are given if the child has received fewer than three previous doses or if the immunization history is unknown; tetanus toxoid should be given if it has been more than 5 years since the most recent booster.

Clinicians should be aware of the risk of rabies virus transmission in children exposed to “potentially rabid” animals, including dogs (uncommon in the United States), bats, foxes, skunks, and raccoons.³⁸ Prophylaxis against rabies virus should be considered for any wound (bite or nonbite) resulting from a raccoon, skunk, bat, or fox. Rabies immune globulin should be injected into the wound; the vaccine should be administered in all patients with suspected exposure.³⁸

In general, younger children are less able to communicate information about their injury and pain. Children are more likely to experience an adverse reaction to a product, because allergies are often undiagnosed earlier in life. Scars in children often remain pink and raised for a longer period than in adults; this should be communicated early and frequently to the patient and family to assuage anxiety regarding cosmesis and overall healing.

Wound Care Techniques

Particular techniques for wound care should be chosen based on the time course, location, severity, and mechanism of the wound. Bleeding should initially be controlled with pressure, packing, ligation, or cautery.¹ Pain and anxiety management is essential and may necessitate the use of intravenous opioids and/or anxiolytics. An effective regimen for moderate sedation in younger children is 0.02 to 0.05 mg/kg of midazolam and 0.2 to 0.75 mg/kg of ketamine. When possible, sedation should be performed by a certified specialist who is not the surgeon or wound consultant. Local anesthetics are effective, and additional doses can be given if needed; well-known maximum safe doses of lidocaine are 5 mg/kg without added epinephrine and 7 mg/kg with epinephrine.¹ Distraction techniques are recommended (for example, sucrose for babies, bubbles, lights, and electronic devices), and collaboration with a child life specialist, when available, is very valuable for the patient, parents, and surgeon performing the procedure.² General anesthesia may be required.

Thorough irrigation and debridement are required to cleanse and assess the extent of the wound, remove devitalized tissue, optimize healing, and prevent traumatic tattooing.³⁵ Irrigation should be performed with warm sterile water or normal saline solution.² Again, adequate sedation and analgesia are essential to minimize fear and discomfort, particularly in young children. Primary closure should be attempted whenever the wound is clean, viable, and has little tension. In children, the use of adhesive tapes (Steri-Strips) and skin glue can minimize the need for placing sutures. In infants and toddlers it is particularly important to use absorbable suture material that does not require removal. Either fast-absorbing chromic gut or 7-0 Vicryl is appropriate and will absorb quickly. When possible, wounds on the face should be closed with absorbable suture in children of all ages.

Vacuum-assisted wound closure has many applications and may be used in children.³⁹ Although its use has not been extensively studied in the pediatric population, several initial studies suggest that NPWT is safe and effective in children.³⁹⁻⁴⁴ NPWT offers the benefits of less frequent dressing changes, a clean wound environment, a compact system that facilitates return to activities of daily living, and often less pain, less edema, and possibly fewer operations with less extensive reconstruction.⁴² Multiple studies have estimated that NPWT may also be cost-effective compared with traditional dressing changes (the difference in cost is likely attributable to the cost of long-term nursing care).^{41,43} The literature varies with respect to the recommended pressure

setting: Naik et al⁴⁰ suggest the use of -50 to -100 mm Hg in infants, -75 to -125 mm Hg in toddlers and younger children, and up to -150 mm Hg in adolescents (as in adults). A setting as low as -20 mm Hg may be appropriate for neonates.⁴⁵ To minimize pain while the vacuum dressing is in place, continuous mode rather than intermittent mode should be used. Children may form granulation tissue at a faster rate than adults, and thus dressings may need to be changed more frequently (for example, every 24 hours).³⁵

Wound Care Dressings and Products

A plethora of dressings, topical agents, and wound care adjuncts is available for use with pediatric patients. The selection of dressings and topical agents should be individualized and reflect the characteristics of the wound in question, such as the presence or absence of infection, necrosis, exudate, moisture, damage to surrounding skin, granulation tissue, and exposed underlying structures.³ Dry and wet-to-dry gauze dressing changes have long been used for serial debridement and wound cleansing. However, the use of these dressings can be painful, especially for children. Many additional products, such as transparent films, nonadherent gauzes, hydrocolloids, hydrogels, hydrofibers, alginates, and foams, have occlusive and semiocclusive properties that provide moisture, remove exudate when necessary, minimize bacterial burden, and/or minimize pain. A comprehensive review of available products is beyond the scope of this chapter, but several referenced articles offer more detailed descriptions and evidence-based recommendations for clinical practice.^{2,3,35,46,47}

Medical grade honey is thought to be safe for use in pediatric patients, although rigorous efficacy studies in neonates and children are relatively limited.³ A Cochrane Review⁴⁶ that included 25 trials of the use of honey-containing products in acute and chronic wounds concluded that honey “might be superior to some conventional dressing materials,” although more research is required. Few studies have evaluated the efficacy of honey-based wound products in pediatric patients. Simon et al⁴⁸ reported their experience with Medihoney in 14 pediatric oncology patients; they stated that topical honey is a useful adjunct for wound healing in patients with immunosuppression.

Dressings and topical products containing silver (specifically nanocrystalline silver dressings, such as Acticoat) may be safe for use in neonates and children when clinically appropriate.^{3,49,50} However, there is concern about the possibility of silver toxicity, and more studies are needed to evaluate the effects of silver-containing dressings in children.^{2,3}

A systematic review of “modern” dressings and topical agents for acute and chronic wounds included 99 studies and found that hydrocolloids and foam dressings are probably efficacious in chronic wounds, and there is evidence to support the use of hydrofibers in acute wounds.⁴⁷ More rigorous scientific and clinical research is needed to better determine recommendations for the use of products containing silver, honey, HA, and activated charcoal, particularly in neonates, infants, and children.

Regardless of the method of wound closure, the use of skin barriers and barrier dressing, cream, or paste should be considered. These products minimize pain with dressing changes and reduce the risk of infection.³⁵ In addition, latex-free and hypoallergenic products should be used when possible.³⁵

Posttreatment Considerations and Complications

Regardless of the chosen method of wound closure or wound care, every effort should be made to keep the skin edges and sutures clean. An overlying dressing should remain in place for 24 to 48 hours to facilitate initial epithelialization. Wound infections may occur, necessitating the use of oral (and sometimes intravenous) antibiotics. Infants and children may develop hypergranulation in response to a wound, whereby granulation tissue grows beyond the boundaries of the

previous injury. This prevents epithelialization, may result in bleeding, and can be painful.³⁵ Topical triamcinolone cream for 1 to 2 weeks can be attempted to minimize hypergranulation.³⁵

Several techniques and products have been discussed to promote optimal scar healing. Sun protection is essential. Scar massage is easily taught to both children and parents; this involves applying pressure to soften scar tissue. Topical ointments and silicone sheeting can provide moisture and warmth to the evolving scar. A relatively new, adherent, silicone-based product, the Neodyne embrace Advanced Scar Therapy device, is applied to incisional wounds; it has been shown to improve the appearance of scars after abdominoplasty.⁵¹ Hypertrophic scar or keloid formation may occur in children, often requiring a multimodal approach to minimize irritation and improve cosmesis. Options include steroid injections, pressure garments, and excision, as discussed earlier in this chapter.

FUTURE DIRECTIONS

With respect to fetal healing, pediatric healing, and regenerative medicine, much has been discovered to date, but more basic science and rigorous clinical research is needed. Advances in tissue engineering, a better understanding of the role of stem cells, and the development of “ideal” biologic substitutes could revolutionize pediatric wound healing. A better understanding of the process of fetal healing may promote the development of improved techniques and indications for fetal surgery and may also facilitate the development of novel ways to promote scarless healing in children and adults.

KEY POINTS

- A thorough understanding of normal and abnormal wound healing mechanisms is important for optimal wound assessment and management in infants and children.
- The diagnosis of, differentiation between, and treatment algorithm for hypertrophic scars and keloids is an essential skill for any physician working with children.
- Compared with healing in adults, fetal healing is characterized by decreased inflammation, a more loosely organized and type III collagen, and different concentrations of ECM components (for example, more HA).
- An enhanced understanding of the dynamics of fetal wound healing will someday allow scientists and clinicians to replicate key fetal mechanisms in children and adults.
- Absorbable suture is recommended for wound closure in infants and young children.
- Perioperative wound care can optimize healing following many surgical interventions.

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7

Fetal Surgery

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Fetal diagnosis facilitated by imaging and fetal sampling has improved tremendously since the mid-1970s, allowing earlier and more accurate diagnosis of fetal anomalies. Surgeons are increasingly involved in the surgical management of these anomalies before birth. Most of these conditions are best managed expectantly, with definitive treatment after birth. However, some prenatally diagnosed conditions are progressive and can result in fetal death or threaten the well-being of the mother. Although some fetal therapies have resulted in great success, many others have shown little or no benefit.^{1,2} The current applications of fetal surgery are outlined in Table 7-1. Almost all fetal therapies are directed at lethal conditions, the most notable exception being myelomeningocele.

FETAL DIAGNOSIS

Fetal ultrasonography is the mainstay of prenatal imaging. Most anatomic surveys are conducted in the middle of the second trimester between 18 and 20 weeks' gestation. The accuracy of fetal ultrasonography depends on the skill and experience of the ultrasonographer and radiologist. Real-time fetal ultrasound identifies anatomic anomalies, provides physiologic information such as fetal heart rate, placental blood flow, and the presence or absence of fetal hydrops, and is critical for guidance during invasive fetal interventions. MRI is increasingly used when high-resolution fetal imaging is needed for the brain, spine, and body. Many syndromes are associated with congenital heart defects; thus fetal echocardiography is also an important diagnostic modality. The presence of a congenital heart defect precludes most fetal interventions.

Table 7-1 Potential Applications of Fetal Surgery

Indication	Anatomic Anomaly	Pathophysiologic Effects(s)	Treatment
Placental Vascular Anomalies			
Twin-twin transfusion syndrome	Unequal placental sharing; abnormal vascular connections	Fetal hydrops/death Surviving twin with severe morbidity	Fetoscopic laser ablation
Twin reversed arterial perfusion syndrome	Acardiac cotwin relies on normal cotwin heart for viability	High-output cardiac failure/hydrops	Selective reduction with RFA or umbilical cord ligation
Urologic			
Fetal obstruction uropathy	Lower urinary tract obstruction (e.g., posterior urethral valves)	Renal failure Pulmonary hypoplasia	Vesicoamniotic shunt
Thoracic			
CDH	Hole in the developing diaphragm	Pulmonary hypoplasia and hypertension	Tracheal occlusion
CPAM and pulmonary sequestration	Space-occupying lobar mass	Low-output cardiac failure Fetal hydrops/death	Maternal steroids Thoracoamniotic shunt
Tension hydrothorax	Lung hypoplasia	Pulmonary failure	Fetal lobectomy
Congenital high airway obstruction syndrome	Complete tracheal or laryngeal obstruction	Low-output cardiac failure/hydrops	Thoracoamniotic shunt Fetal tracheostomy or early delivery through the EXIT procedure
Cardiac			
Pericardial teratoma	Tumor mass compressing heart and great vessels	Fetal hydrops/death	Fetal sternotomy and resection
Pulmonary/aortic stenosis	Obstructed valves	Heart failure Single-ventricle physiology	Catheter-based balloon dilation
Ebstein's anomaly	Pulmonary compression from atriomegaly Pulmonic valvular atresia Tricuspid regurgitation	Fetal hydrops/death Pulmonary failure	Atrial reduction, valve repair
Neurologic/Posterior			
Myelomeningocele	Notochord malformation Chiari malformation	Hydrocephalus Neurogenic bladder/bowel Orthopedic anomalies Somatosensory loss	Fetal repair Fetal resection
SCT	Vascular "steal" through tumor mass	High-output cardiac failure/hydrops	

CDH, Congenital diaphragmatic hernia; *CPAM*, congenital pulmonary airway malformation; *EXIT*, ex utero intrapartum treatment; *RFA*, radiofrequency ablation; *SCT*, sacrococcygeal teratoma.

Biochemical screening is an important noninvasive diagnostic tool for evaluating aneuploidy. Noninvasive genetic testing for fetal disease is now possible with powerful sorting techniques that allow isolation of fetal cells and free fetal DNA from the maternal circulation.³ Fetal sampling remains an important method of karyotyping and DNA-based diagnosis of many genetic defects and inherited diseases. Amniocentesis provides a safe method of fetal sampling, but it cannot be employed until the second trimester. Chorionic villus sampling is increasingly being utilized, because it can be employed as early as 10 weeks of gestation.

FETAL ACCESS

The technical aspects of safely accessing the fetus for intervention evolved over 30 years of experimental and clinical work. In general, there are three methods for accessing the fetus:

1. Open hysterotomy
2. Minimally invasive fetoscopy
3. Percutaneous access

All three approaches require preoperative and intraoperative ultrasonography to define the anatomy, determine the position of the fetus, delineate the placental anatomy, and monitor fetal heart rate and umbilical artery blood flow during the operation.

The fetal surgery team generally consists of two pediatric surgeons, a maternal fetal medicine specialist, an ultrasonographer, and an obstetric anesthesiologist. The mother is supine with the left side down to minimize compression of the inferior vena cava. Depending on the nature of the intervention, maternal anesthesia can be either spinal or general. When needed, fetal anesthesia is administered through an intramuscular injection of an opiate and a nondepolarizing muscle relaxant.

Open fetal surgery requires general anesthesia, preoperative indomethacin, and high-dose inhalational agents to maintain uterine relaxation. Fig. 7-1 provides a summary of the open fetal surgery technique. The uterus is exposed through a low transverse abdominal incision. Ultrasonography is used to map out the placenta. A posteriorly positioned placenta allows an anterior hysterotomy. An anteriorly positioned placenta requires elevation of the uterus from the abdomen through a posterior hysterotomy. The hysterotomy is made with specially designed absorbable uterine staples that provide hemostasis and seal the membranes. The fetus is monitored with continuous pulse oximetry and echocardiography. Warm saline solution is continuously infused around the fetus to prevent hypothermia and compression of the cord vessels. Fetal exposure is limited to the body part of interest, and care is taken to prevent herniation of the fetus. After completion of the fetal operation, the fetus is returned to the uterus, the amniotic fluid is replaced, and the hysterotomy is closed with running and interrupted absorbable sutures. Postoperatively, tocolytics are administered and the mother is monitored for contractions. The fetal heart rate is monitored and daily echocardiography is used to monitor for ductus arteriosus constriction and right-sided heart failure.

Minimally invasive fetoscopic surgery is usually performed through a 3 mm fetoscope with a 1 mm working channel. Intraoperative real-time ultrasonography is critically important for identifying a safe point of access on the uterus that is free of large vessels and placental attachment. Furthermore, ultrasonography is an indispensable adjunct to fetoscopic visualization. Clinical applications of fetoscopic surgery include laser ablation of placental vessels in twin-twin transfusion syndrome, fetal cystoscopy and urinary tract decompression, division of amniotic bands, and tracheal occlusion for *congenital diaphragmatic hernia* (CDH). Percutaneous ultrasonographically guided interventions include catheter shunt placement (bladder and chest), radiofrequency ablation (RFA) of large tumors and anomalous twins, aspiration of fluid from fetal body cavities, and administration of drugs or cells directly to the fetus.

The *ex utero intrapartum treatment* (EXIT) procedure was developed as a bridge between fetal and postnatal therapy. Anesthesia is delivered to both the mother and fetus, and a hemostatic hysterotomy is made. The uterus is completely relaxed to prevent placental separation, and the umbilical cord is not clamped. In this way, the fetal circulation is preserved and the fetus is maintained on full placental support until the airway can be secured. The EXIT procedure has been used to reverse tracheal occlusion, repair the trachea, place a tracheostomy tube, resect large cervical tumors (Fig. 7-2), and to place cannulas for immediate *extracorporeal membrane oxygenation* (ECMO).⁴

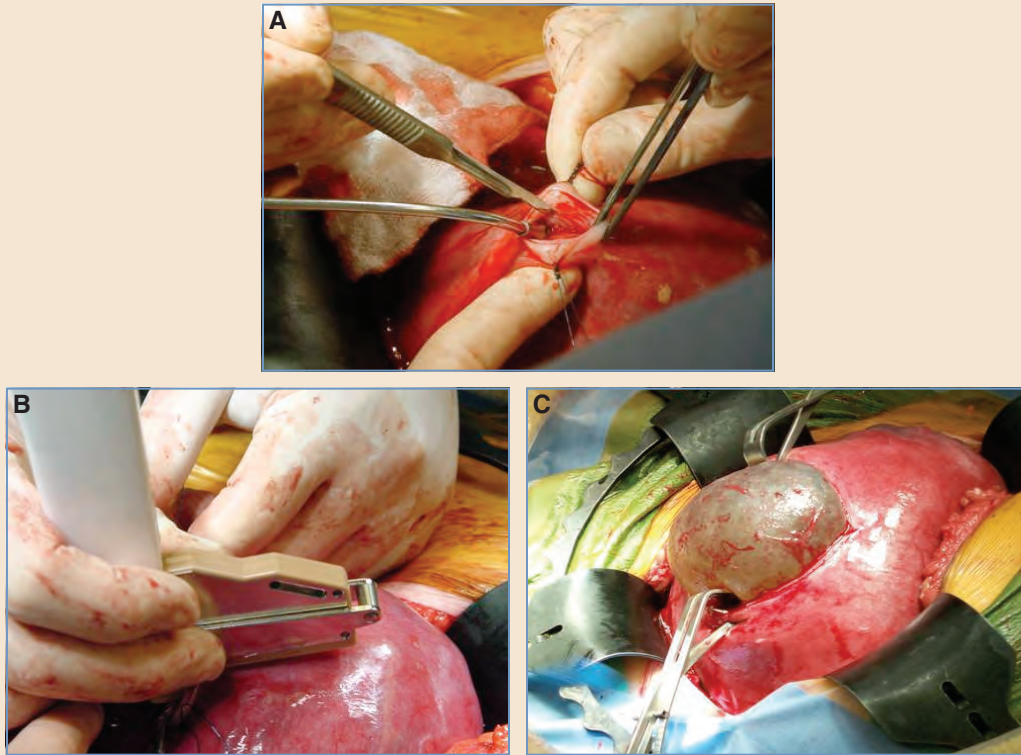


Fig. 7-1 A, A hysteroscopy is performed using a scalpel and suction. An opening of approximately 2 cm is created sharply. If possible, the dissection is carried down to, but not through, the membranes. B, The uterine stapler is applied. If kept extraamniotic, the risk of injuring a fetal part with the stapler is minimized. Two rows of hemostatic absorbable staples are applied, and the myometrium is cut between the two rows. C, An “amniotic bubble” appears after the myometrium is incised. Note the excellent hemostasis and the two uterine retractors. The membranes can now be opened sharply with scissors. An amniotic bubble cannot be created in all cases. Often the uterine cavity is opened at once. This is not a disadvantage, although extra care must be exercised to avoid injuring a fetal part or the umbilical cord when the stapler is applied.

Fig. 7-2 This fetus underwent delivery using the EXIT strategy. Note the large cervical teratoma that, on rigid bronchoscopy, was completely obstructing the trachea. A resection was performed while the fetus was on “placental support.” After the teratoma was excised, the trachea was intubated transorally, the umbilical cord was cut, and the fetus was “officially” delivered.



MATERNAL RISK

Fetal intervention places both the unborn patient and mother at risk. The mother gains nothing in terms of personal health benefits, but she is placed at significant risk for morbidity and potential mortality. Since the first open fetal surgery at the University of California, San Francisco in 1982, there have been no reported maternal mortalities.⁵ Despite technical advances in fetal intervention, disruption of the membranes and preterm labor are the Achilles heel of fetal surgery, resulting in premature delivery at approximately 34 weeks' gestation. Although fetoscopic techniques might theoretically mitigate maternal risk, the results of multiport fetoscopy have been disappointing, mostly because of membrane problems leading to preterm labor. The most common immediate maternal complication is pulmonary edema associated with the administration of tocolytics. Bleeding that requires transfusion is an infrequent but significant problem. Other complications include infection in the form of chorioamnionitis, pulmonary embolism, and deep venous thrombosis.⁴ Importantly, fetal surgery does not appear to diminish fertility or the ability to carry future pregnancies. However, the delivery of pregnancies after fetal surgery requires cesarean section because of the risk of uterine rupture.⁶

ANOMALIES AMENABLE TO FETAL SURGERY

Congenital Diaphragmatic Hernia

Congenital diaphragmatic hernia (CDH) interferes with normal lung development and is associated with 30% to 40% mortality as a result of pulmonary hypoplasia and pulmonary hypertension. Fetal ultrasound is used to stratify the severity of CDH. Liver herniation into the chest is the most important prognostic factor for isolated CDH, and is associated with markedly increased mortality.⁷ The *lung-to-head ratio* (LHR) was developed to further stratify the severity of CDH relative to lung size. The LHR compares lung size to head circumference to control for differences in gestational age and size. A fetus with liver herniation and an LHR of less than 0.9 has a very poor prognosis and is a candidate for fetal intervention.⁸

We hypothesized that in utero correction of the CDH and reduction of herniated abdominal viscera from the chest would allow antenatal lung growth and improved survival. Initial attempts at fetal surgery to treat CDH sought to correct the anatomic diaphragmatic defect in utero. Although CDH repair through open hysterotomy proved feasible, there was no survival benefit and this strategy was abandoned.⁹ Efforts were redirected at finding a strategy that promoted in utero lung growth. The observation that fetuses affected by congenital high airway obstruction developed hyperplastic lungs led to a series of animal experiments demonstrating that fetal tracheal occlusion stimulates lung growth. In 2003 the National Institutes of Health (NIH) funded a trial comparing in utero fetoscopic tracheal occlusion with standard postnatal care in fetuses with severe CDH.¹⁰ The tracheal occlusion group required an EXIT procedure at birth to remove the tracheal balloon. Although the tracheal occlusion group met survival expectations (73%) that were better than historical controls (37%), tracheal occlusion proved no better than standard therapy in the concurrent control group (77%).¹⁰

Fetal tracheal occlusion techniques continue to evolve. Temporary tracheal occlusion improves fetal lung volume, whereas long-term tracheal occlusion can be deleterious to type II pneumocytes. Temporary fetal tracheal occlusion can now be achieved with a tracheal balloon that is both deployed and later retrieved, using a percutaneous fetoscopic technique (Fig. 7-3). In utero removal of the tracheal balloon obviates the need for an EXIT procedure. The European experience with temporary fetal tracheal occlusion for severe CDH has been favorable, with survival of 50%. However, the dismal survival rate of 15% in the standard therapy group

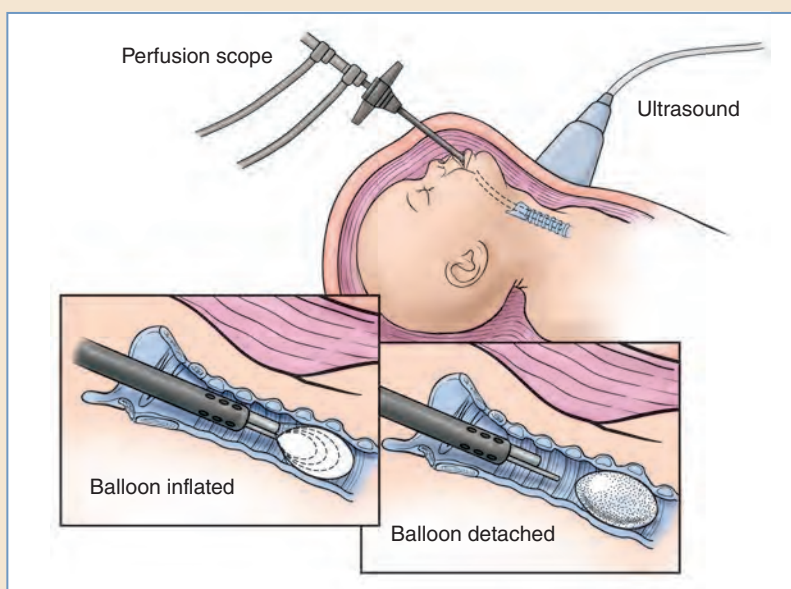


Fig. 7-3 Using a single trocar, a perfusion endoscope with a working channel is inserted into the fetal mouth, and direct laryngoscopy is performed. After placing the endoscope into the midtrachea (facilitated by ultrasonographic guidance), a detachable balloon is deployed using a small catheter through the endoscopic working channel.

has come under criticism.¹¹ A prospective randomized trial of temporary fetal tracheal occlusion versus standard therapy is under way in Europe, and safety and feasibility testing of the device are ongoing in the United States.

Tumors

Fetal tumors are rare, and the vast majority of them are best dealt with postnatally. However, very large tumors can lead to fetal hydrops that results in fetal demise and thus they must be addressed in utero. Tumors cause nonimmune fetal hydrops by compressing the vena cava and compromising preload of the heart or from high-output cardiac failure through arteriovenous shunts within the tumor. Hydropic changes include pericardial effusion, pleural effusion, abdominal ascites, and scalp edema, as well as polyhydramnios and placental megaly.¹²

Congenital pulmonary airway malformation (CPAM), previously referred to as *congenital cystic adenomatoid malformation (CCAM)*, is characterized by an overgrowth of the respiratory bronchioles with associated cyst formation. Although most CPAMs can be managed postnatally, very large CPAMs can result in fetal hydrops that rapidly progresses to fetal death. The size of the CCAM is the most important risk factor for developing fetal hydrops. The *CCAM volume ratio (CVR)*, which describes the proportion of CCAM volume to head circumference, has been shown to be predictive of fetal hydrops.¹³ Microcystic CPAMs tend to grow until 26 to 28 weeks' gestation and then plateau or regress in size, whereas macrocystic lesions generally continue to grow throughout gestation. Fetal pulmonary lobectomy for CPAM with associated hydrops has been shown to reverse fetal hydrops, but it is associated with preterm labor and prematurity.¹⁴ Interestingly, mothers who were given steroids in preparation for fetal pulmonary lobectomy and who

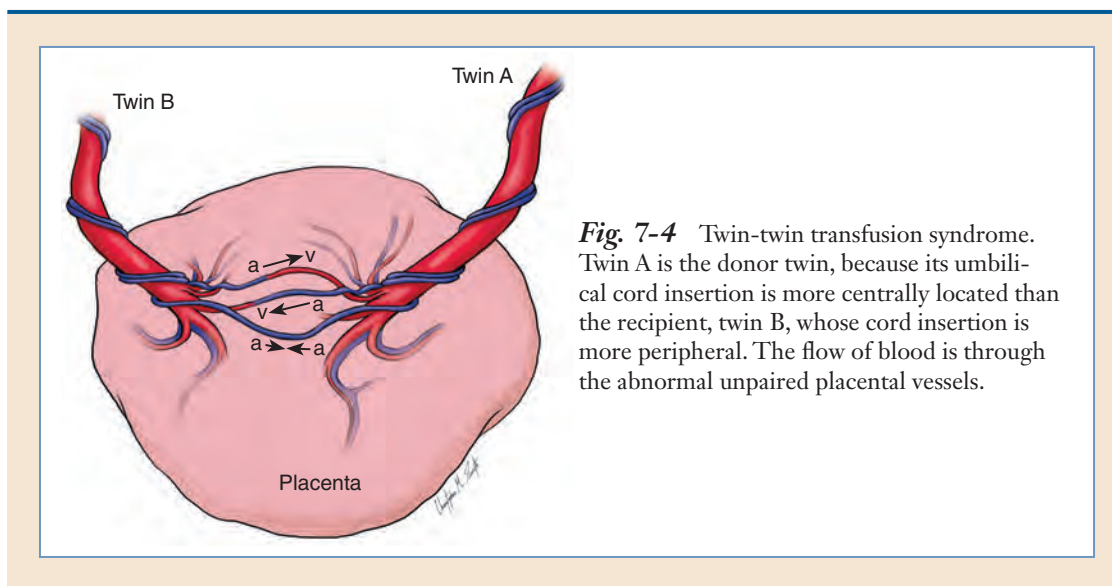


Fig. 7-4 Twin-twin transfusion syndrome. Twin A is the donor twin, because its umbilical cord insertion is more centrally located than the recipient, twin B, whose cord insertion is more peripheral. The flow of blood is through the abnormal unpaired placental vessels.

then did not undergo the operation showed regression of the hydrops.¹⁵ Subsequent prospective studies showed that maternal administration of corticosteroids for large microcystic CPAMs is highly effective at reversing hydrops, whereas macrocystic CPAMs do not respond to corticosteroid administration.¹⁶ Macrocystic CPAMs with associated hydrops are best treated with percutaneous thoracoamniotic shunt placement.¹⁷ Fetal pulmonary lobectomy is now reserved for the rare patients in whom corticosteroids or thoracoamniotic shunting has failed.

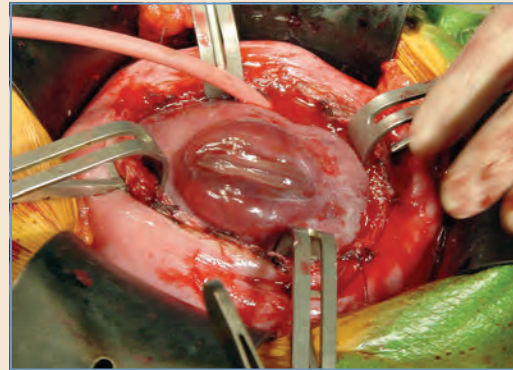
Sacroccygeal teratoma (SCT) is another tumor that is commonly diagnosed prenatally. As with CPAM, SCT can result in fetal hydrops and death. SCTs can grow rapidly to an enormous size relative to the fetus. Attempts at ablating the tumor, either through alcohol injection or RFA, have proven unsuccessful. Fetal surgery to debulk the large external component of an SCT with associated hydrops has proven successful.¹⁸

Abnormalities of Twin Gestations

Monochorionic twins, identical twins that share a single placenta, can have problems with unequal blood flow or unequal sharing of the placenta. *Twin-twin transfusion syndrome* (TTTS) occurs in 10% of monochorionic pregnancies, and is the result of net blood flow from one twin to the other (Fig. 7-4). The “recipient” twin gets too much blood and is at risk of high-output cardiac failure, whereas the “donor” twin gets too little blood flow and is at risk of hypovolemia and hypoperfusion. TTTS carries a 90% mortality rate for both twins if left untreated. Fetoscopic laser ablation of the abnormal vascular communications between these twins has become the benchmark treatment for TTTS, with single and dual survivorship of 76% and 36%, respectively.^{19,20}

Twin reversed arterial perfusion (TRAP) *sequence* is a rare anomaly of monochorionic twins in which a normal twin acts as the “pump” for an acardiac, acephalic twin. As the acardiac, acephalic twin grows in size, the pump twin goes into high-output cardiac failure and hydrops. If left untreated, TRAP is associated with 50% fetal demise of the pump twin. Percutaneous ultrasound-guided RFA of the umbilical cord as it enters the abdomen of the acardiac twin has proved to be highly effective at halting TRAP and protecting the pump twin, with an overall survival of 80%.^{21,22}

Fig. 7-5 Intraoperative surgery for a fetal thoracolumbar myelomeningocele. Note the four hysterotomy retractors and the catheter that provides continuous warmed irrigation fluid to the fetus during the operation.



Myelomeningocele

Myelomeningocele (MMC), or *spina bifida*, is a devastating birth defect resulting from an open neural tube and exposed spinal cord. The rationale for in utero repair of MMC is that the exposed spinal cord becomes secondarily damaged by amniotic fluid exposure, direct trauma, and hydrodynamic pressure. Various animal models demonstrated that in utero repair of MMC prevents this secondary injury and results in improved neurological function.^{23,24}

MMC is the first nonlethal anomaly for which fetal surgery was undertaken.²⁵ Early attempts at fetal MMC repair in humans showed promise, but it was difficult to know the true benefit when compared with historical controls. The NIH funded a multicenter randomized trial (Management of Myelomeningocele Study [MOMS])²⁶ comparing fetal MMC repair at 19 to 26 weeks' gestation with postnatal repair (Fig. 7-5). The study ended early because of the superiority of fetal surgery. Fetal MMC repair reduced the need for ventricular shunting for hydrocephalus at 12 months (40% for the fetal group versus 82% for the postnatal group, $p < 0.001$) and improved motor function, including the ability to walk at 30 months of age (42% for the fetal group versus 21% for the postnatal group ($p < 0.01$)).²⁶

IN UTERO STEM CELL TRANSPLANTATION FOR GENETIC DEFECTS

With the advent of chorionic villus sampling, various inherited genetic defects can be detected early in gestation. A number of these defects, such as immunodeficiencies, hemoglobinopathies, and storage diseases, are potentially curable with in utero hematopoietic stem cell transplantation.²⁷ Before 15 weeks' gestation, the fetus is in a preimmune state, and the bone marrow is primed to receive hematopoietic stem cells migrating from the liver. Thus in utero transplantation may avoid problems with rejection, tolerance, and graft-versus-host-disease that occur with postnatal transplantation.²⁸ However, delivery of even a small volume of cells to the fetus early in gestation is technically demanding and carries significant risk. Furthermore, it is unclear whether a sufficiently high degree of engraftment can be achieved for a cure. Although transplantation for severe combined immunodeficiency syndrome has been successful, the promise of in utero stem cell therapy for other disease has yet to be fulfilled.

PLASTIC SURGERY AND FETAL INTERVENTION

Early intervention for congenital deformities of the head and neck or limbs has similar appeal to in utero stem cell treatment: the potential to reverse secondary deformities by correcting the

inciting problem. The finding that the fetus heals without visible or histologic scarring before approximately the middle of pregnancy makes the repair of cleft lips and other congenital deformities particularly attractive.

Scarless Healing

Early in the process of developing fetal surgery, the observation was made that the fetus heals without scarring up to the middle of the second trimester, which is roughly the middle of the pregnancy. This is obviously intriguing, because a cleft lip, for example, might be repaired leaving no visible scar. Improved prenatal ultrasonography allows the diagnosis of clefts and other craniofacial anomalies by 20 weeks' gestation, which would be early enough for scarless healing. A better understanding of the mechanism of scarless healing might also allow alteration of the wound healing milieu in the postfetal environment to improve or even eradicate scar. Despite extensive investigation into the nature of fetal wound healing, the process remains poorly characterized.²⁹ Many differences in specific markers have been elucidated between the fetal and adult wound-healing environments,³⁰ but attempts to induce fetal healing in the adult environment have been generally unsuccessful. It appears that the ability of fetal wounds to heal without scarring is intrinsic to the fetal epithelial cell rather than the amniotic environment, making it more difficult to apply to the adult situation.

Cleft Lip And Palate

Intrauterine cleft lip repairs have been performed in both large and small animals. Smaller animals such as rat and rabbit models allow inexpensive interventions, but because of their very short gestation, these can be seen primarily as proof of concept.^{31,32} The primary model for fetal cleft lip repair has been in sheep; there is not a reliable large animal model for clefts, necessitating surgical creation of the cleft lip and alveolus. In addition, sheep have proved to be tolerant of intrauterine interventions without the preterm labor that is precipitated in primates, including humans.

Initial efforts at fetal cleft repair used an incisional model, performing repair at the same time.³³ Later a two-procedure model was used to more closely simulate the clinical situation, creating the cleft surgically at approximately 60 days gestation, and then repairing the cleft at a second procedure at approximately 90 to 100 days.³⁴ A subsequent investigation compared neonatal repair with in utero repair in the long-term recovery of fetal lambs (6 months). This showed that both groups had some lip shortening that would require secondary surgery (and therefore an adult-type scar); however, the group repaired in utero did not have maxillary growth inhibition, which was a significant and unfavorable finding in the postnatal repair group.³⁵

The development of endoscopic techniques for fetal intervention (also known by the term coined by Harrison, *fetendo*, because of the similarity of its manipulation to a video game) was performed in the hope of decreasing preterm labor, although this has not been the result, as previously mentioned. Endoscopic repair of cleft lips a number of years ago was limited by the small space for maneuvering and the large instruments designed for laparoscopic surgery.³⁶ In the future, robotics with multiple degrees of freedom at the end of the instrument may allow much more accurate endoscopic intervention. Alternatively, better control of preterm labor with tocolytics may facilitate more open intrauterine procedures.

Craniosynostosis

Craniosynostosis of either single or multiple sutures is generally best treated early. There is a large body of evidence that strip craniectomy alone is effective if performed early enough. We created a fetal model for unilateral coronal craniosynostosis by placing a mixture of bone

morphogenetic protein (BMP) and TGF-beta into a cranial defect in a fetal lamb. This produced a phenotype analogous to that in humans, with flattening of the frontal region on the affected side and compensatory bossing on the contralateral side.³⁷ In a subsequent study we returned after 3 to 4 weeks to perform strip craniectomy of the stenosed side and showed that the phenotypic findings were all reversed.³⁸

Although fetal intervention for craniosynostosis may not be currently practical in humans, these articles indicate that it may eventually be possible to perform an earlier minimal intervention that will reverse some of the late findings seen in these complex craniofacial anomalies.

Amniotic Bands

Amniotic bands have been implicated in limb abnormalities ranging from indentations in the extremities to intrauterine amputations. In addition, they have been proposed as a cause of orofacial clefts.

Experimental work, again in sheep, showed that a suture or umbilical tape around an extremity could produce an amniotic band “type” outcome, and that early release of the band would reverse those findings.³⁹ In another study our group demonstrated orofacial clefting, similar to that found in humans when a tie was placed intraorally to the lateral face.⁴⁰

The fact that a severe deformity can result from a single band led to the conclusion that a simple endoscopic release of the band could reverse the outcome. This has now been accomplished in numerous people with good results.^{41,42}

KEY POINTS

- Most congenital anomalies are managed expectantly during pregnancy, with definitive treatment after birth.
- Fetal surgery is largely reserved for fetuses with lethal anomalies, the notable exception being fetal MMC repair.
- Experienced multidisciplinary teams are essential for the success of any fetal therapy program.
- NIH-funded prospective randomized trials have shown the benefit of fetal surgery in MMC and TTTS.
- The future of fetal therapy lies in stem cell and genetic therapies for the correction of genetic defects in utero.
- Improved techniques of endoscopic intervention and control of preterm labor after fetal intervention may permit broader application for congenital deformities.

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8

Cutaneous Lesions

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utaneous lesions are common in children and are a source of concern for parents, pediatricians, and surgeons alike. Fortunately, most of these lesions are benign. Despite this fact, many patients seek surgical removal of the lesions. For parents and older children, the visibility of the lesion and teasing by peers are frequently factors that prompt a desire for removal. In particular, fear of future malignant degeneration is a motivating force among families and physicians alike. The role of the pediatric plastic surgeon is to educate the family and referring physician about the nature of the lesion so they can make an informed decision regarding the need for removal, balancing this with appropriate expectations for the scar that will follow. If excision is recommended, the pediatric plastic surgeon must suggest the best timing with respect to the natural history of the lesion, the physical and social development of the child, and how the operative site and reconstructive plan might be affected by subsequent growth. Finally, the plastic surgeon should remove the lesion with appropriate margins for the type of lesion and in the most aesthetically pleasing manner possible.

In this chapter we will discuss the issues that must be considered when excising skin lesions in the pediatric population and focuses on the specifics of commonly occurring cutaneous and subcutaneous lesions. The discussion is divided into melanotic and nonmelanotic lesions. Hypertrophic scars, keloid scars, neurofibromas, and vascular lesions are covered in Chapters 6, 11, and 37.

TIMING OF SURGERY

Most skin lesions in children can be removed electively, leaving the timing of surgery to a mutual decision between family and surgeon. Parents of infants and preschool children with lesions in visible places such as the face, arms, and legs may have concerns about the social stigma for the child and may desire removal of these before he or she reaches school age. Parents concerned about the risks associated with anesthesia may want to defer surgery to a time when removal could be performed with a local anesthetic. Families concerned about malignant degeneration

may have an unwarranted sense of urgency. These family biases should be balanced against the child's ability to cooperate with both the planned procedure in the office and postoperative limitations and care.

Office sedation of children can be difficult because of their size and the need for weight-appropriate doses of sedatives and reversal agents. Office procedures are better conceived of as local anesthesia cases. Other situations requiring sedation are best performed in a surgical center or operating room with the assistance of anesthesia personnel. These limitations mean that most infants and toddlers are not candidates for office procedures. Most cases in which local anesthetic is used should not be attempted until the physician can engage the patient in meaningful conversation. Some children are ready for office surgery at 4 to 5 years of age, whereas others are not ready until much later. The patient's motivation and verbal skills are the best indicators of patient selection for office surgery.

Despite the low acuity of most pediatric cutaneous lesions, the energy and time that can be expended assessing and treating these smaller, benign cases can be quite significant. Parents often come to the pediatric plastic surgeon's office with preconceived notions of what should happen. Families with a history of skin cancer or melanoma often are overly concerned about malignancies in their children. Many come with the misconception that plastic surgeons can operate without leaving a scar or hold hope that lasers and other types of treatment will remove the scar secondarily. The surgeon must realistically describe the balance of a scar versus the visibility of the lesion and encourage the parents to be realistic about the perceptions of a scar versus the lesion in the child's later interaction with peers. Parental guilt and anxiety about scars, visible lesions, or previous medical care can be palpable at times.

The surgeon must also be sensitive to how each child reacts to the possibility of having surgery awake versus being asleep. If the child is clearly distressed about being awake during the procedure, the surgeon can allow the family to delay surgery until the child is old enough to be comfortable with surgery in the office or can clearly state that he or she would prefer to be asleep. The surgeon is the advocate for the young child; if the child is clearly uncomfortable having surgery awake, the surgeon should work to prevent the child's being pushed into it by a parent.

A clear conversation with parents and older children is essential, in age-appropriate language and covering all of these points. Expectations for the quality of scars on different body parts should also be discussed in advance with families. Certain areas such as the jawline, shoulders, and presternal chest are more prone to hypertrophy. Scalp wounds will have areas of residual alopecia. Areas over extensor surfaces such as the back and joints tend to spread over time. Skin grafts, even if well done, will have the appearance of a patch and may be associated with regional contour deformities. Prediction of these events is easier than explanation in retrospect. This information should be reinforced with further meetings and possibly teaching sessions with the nursing staff when staged or complex reconstruction, such as with tissue expansion, is considered. If the family's expectations for the procedure differ from the surgeon's, these generally straightforward cases can weigh one down with unhappy patients who have unrealistic demands. It is important to discuss the postoperative course, including activity limitations and the necessity for active scar management and sun protection. Most of these procedures are elective and can be scheduled around family, school, sports, and patient and surgeon schedules.

EXCISIONS: CAVEATS AND SUGGESTIONS

Although most technical details of excising lesions in children are similar to those in adults—planning incisions to lie favorably, taking adequate margins, undermining surrounding tissue to relieve tension, meticulous hemostasis, and exact realignment of the dermal-epidermal junc-

tion—there are several unique situations that may catch a surgeon off guard whose practice is predominantly with adult patients.

Juvenile skin, although elastic, does not have the laxity of adult skin afforded by gravity. Local head, neck, and extremity flaps that are easily achievable in adults are often difficult in children. Secondary pull on adjacent structures must be anticipated before planning local flaps. When direct excision and primary closure are not possible, the surgeon should entertain other strategies, such as staged excision and skin grafting. If local flaps remain the most desirable form for wound closure, regional tissue expansion can provide the needed tissue and diminish secondary deformation, if the flap is rotated or transposed. Direct advancement might distort the adjacent structures or limit later growth. Free tissue transfer can be used for larger lesions, with or without expansion (see Chapter 12 for more details).

Infantile skin is very thin, seemingly paper-thin at times. Generous undermining, even for small lesions, is essential to prevent sutures from pulling through. Reverse cutting needles also seem to reduce laceration of the dermis in subcuticular suturing. The use of precision-tip blades (Beaver-type 6700 and Bard-Parker 15C style) provides increased control over cutting corners of very small excisions in infants and children.

Patient selection and advance planning are the keys to office excisions with the use of local anesthetic in children. Children amenable to in-office surgery are those whom you can engage in conversation. “Talkaesthesia” is critical to leading the child through the procedure. Calm, straightforward parents are also essential. Children who sense parental anxiety will be unable to cooperate. We favor the use of topical anesthetics such as LMX and lidocaine/prilocaine (EMLA) to diminish the pain of the needle stick and injection of the local anesthetic. Using fine-gauge needles (27-gauge or 30-gauge) and buffering with sodium bicarbonate solution (4 parts local anesthetic with epinephrine to 1 part sodium bicarbonate solution) also help to decrease the sting. Comparing the sensation to the “tingling when your hands or feet fall asleep” is an explanation most children and parents can understand. The surgeon should prepare and drape the area widely to provide ample room to work and use operating room–quality instruments.

The surgeon should also place sutures that do not require great effort to remove. We prefer to use buried dissolvable sutures, and the senior author (BSB) prefers to use pull-out Prolene sutures. We discourage the use of chromic sutures in cosmetically sensitive areas; they do not dissolve rapidly enough to avoid epithelialized tracks. Furthermore, the erythema and induration induced by the cellular mechanism of suture resorption can create concern about wound infection. However, chromic sutures are excellent for all skin repairs on the ears and on the palms and soles, where epithelial and suture tracks seem not to be an issue.

The surgeon should keep dressings simple—Steri-Strips or Dermabond. Acetaminophen (Tylenol), ibuprofen (Motrin), and ice packs should help with postoperative discomfort. The need for postoperative analgesia seems to diminish significantly when either a regional nerve block or periincisional injection is performed with bupivacaine (Marcaine); 0.25% is injected before completion of the procedure. Management of postoperative pain expectations by honest explanations in age-appropriate language is helpful to the patient and parent. For small excisions in the office, we compare the postoperative expectation to a cut from a fall at the park—an event that most children know they have survived.

Postoperative wound management can be challenging in this patient population. Patient and parent education about the natural history and duration of scar maturation can diminish concern about the considerable period of palpable healing scar ridge and pink discoloration (up to 12 to 18 months). Although most parents are willing to perform uncomfortable scar massage on themselves, they often cannot bear to “hurt” their children. Taping with Steri-Strips, Microfoam tape, or over-the-counter silicone sheeting can be helpful. The use of small external dressings for

toddlers and infants should be limited to hours when the parents are watching the child to decrease the risk of the child's removing the bandage and choking on it.

In this era of children with heavily committed schedules of sports and other activities, parents and children, who frequently see missing these activities as catastrophic, must be counseled, often by talking to the child to emphasize that he or she must choose a surgery time that will allow an appropriate break from these activities. It is essential that the surgeon and nursing staff emphasize the importance of limiting activity in the postoperative period to minimize the risk of wound complications and again clarify what is acceptable and what is unacceptable activity. For elective procedures it is often worth delaying surgery until the "off season," even if it means a narrow window of time before the next sports season begins.

MELANOTIC LESIONS

Congenital Melanocytic Nevi

Congenital melanocytic nevi (CMNs) are generally referred to as birthmarks. Although they may be apparent at birth, many gradually become more apparent over the first few years of life as pigmentation increases (tardive appearance). Physical examination reveals that they are plaques, raised from the surrounding skin with increased skin markings and appendages. They can range in color from a tan to a deep blue-black. Neural elements with rugose texture and coarse terminal hair may be present. Surface texture and color may change over time, particularly near puberty, and are not necessarily indicative of malignant change. Some of these lesions will lighten with time and may present with a halo phenomenon during puberty.¹

Embryologically, these tumors are hamartomas consisting of nevus cells, which differ from regular melanocytes by their arrangement into nests or clusters, their rounded dendritic shape, and their tendency to keep their melanin pigment rather than to transfer it to surrounding epithelial cells.² Histologically, the presence of nevus cells within the eccrine ducts and glands, follicular epithelium, or blood vessels is specific for CMNs, but these findings may not be seen in all congenital nevi.³ In larger lesions, nevocytes can be found within subcutaneous fat and, rarely, deeper structures.

CMNs have been classified by their current size or the predicted adult size. In adults, small nevi that have a diameter of 1.5 cm or less are present in approximately 1:100 people. Medium-sized lesions measure between 1.5 and 19.9 cm in diameter and have an occurrence rate of 1:1000. Large lesions are larger than 20 cm and have a reported occurrence rate of 1:20,000 births. A subset of large lesions is the giant nevi, which have a diameter larger than 50 cm and an occurrence rate of 1:500,000 (Fig. 8-1). Although this classification has been used for both prognostic and reconstructive purposes, a more descriptive classification could be more predictive. A new consensus-based classification has been proposed and accepted that includes categories for projected adult size, location, satellite count, and morphologic characteristics (color heterogeneity, rugosity, nodularity, and hypertrichosis).⁴

The reported lifetime risk for melanoma in small and medium CMNs is low, ranging from 0.7% to 2.4%.² Melanoma with small and medium lesions typically occurs postpubertal. Therefore prophylactic removal of small and medium lesions is recommended anytime before puberty, particularly if the lesions are located in areas of irritation (for example, the collar line, belt line, or bra line), if they cannot be easily observed for changes (for example, when on the scalp or the posterior aspect of the body), or if they are in cosmetically sensitive areas. The expected scar must be balanced against the benefits of removal. Small lesions, if favorably located, can be excised and



Fig. 8-1 CMNs occur in a wide variety of sizes, colors, and surface characteristics. **A**, A child with a giant nevus and multiple satellite nevi on his scalp has the variety of small and medium CMNs that present as satellites. **B**, Typical appearance of an isolated medium CMN on the neck. **C**, Medium synchronous or “kissing nevus” of the upper and lower eyelid shows how both the nevus cells migrated into the developing eyelid before its separation, and how a medium nevus may require more than simple excision for treatment. **D**, Congenital giant pigmented nevus with marked variegation in surface color, nodularity, and proliferative nodules.

closed primarily in layers. Medium lesions require more thoughtful reconstruction, including serial excision, grafting, tissue expansion, and local flap coverage (Figs. 8-2 and 8-3).

Even with the low risk of degeneration, these smaller lesions can still be the source of significant peer ridicule, and thus excision may be appropriate before these issues become a major source of psychological distress for the child and family.

Large and giant lesions present more of a management and reconstruction challenge. The immediate concerns are for potential malignancy and the psychosocial development of children with large, obvious lesions. Although the exact occurrence rate is unknown, Tromberg et al³ reported melanoma rates of 4.5% to 10% in large and giant nevi. Older studies reported that 60% of melanomas that arise from large CMNs do so in the first 3 years of life, with 70% occurring by puberty.⁵ When melanoma arises, it may not always be from the nevus itself. Without a prospective, randomized trial, the best evidence for malignancy risk remains population studies.

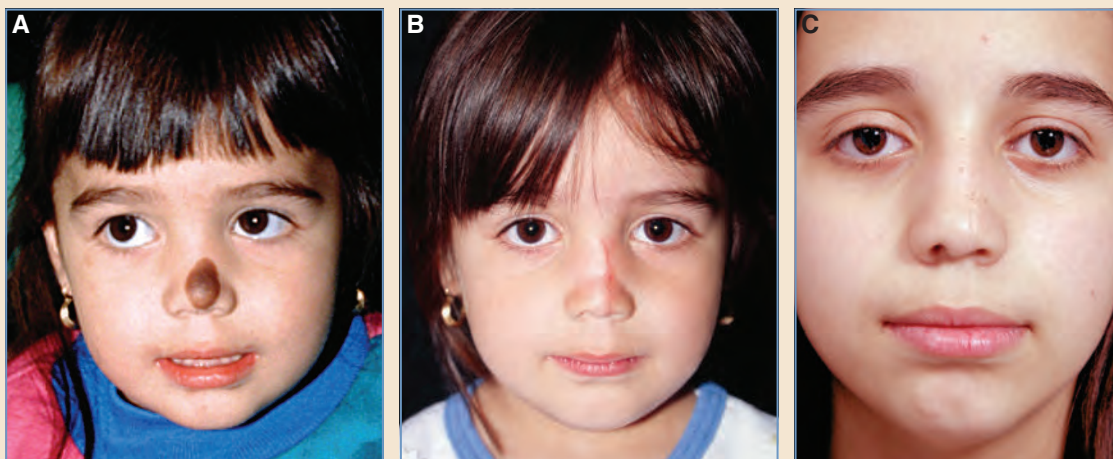


Fig. 8-2 A, This small CMN is in a very visible area of the girl's face. It was treated relatively early, starting at 3½ years of age. B, Appearance after two stages of excision spaced 3 months apart. C, Appearance 12 years after excision.



Fig. 8-3 A, This medium CMN was excised in two stages to minimize tension and overall lengthening of the scar. B, Appearance 1 year after completion of the excision.

A recent Danish population study confirmed the small but real increase in the lifetime risk for developing melanoma in patients with large CMNs.⁴

Further confounding the prognosis of large CMNs is the potential for leptomeningeal involvement, in particular for lesions located over the axial central nervous system. MRI can diagnose the so-called *neurocutaneous melanosis* accurately, but its significance with regard to the development of later melanoma in patients without central nervous system symptoms remains obscure.⁶ However, baseline MRI studies are prudent and can sometimes be predictive of neurologic outcomes.⁷ Understanding that deeper seats of tumor generation likely remain with the patient even after removal of a skin lesion, these surgeries should be considered tumor debulk-

ing rather than complete removal. Periodic lifetime follow-up with these patients will reveal the true nature of the lesion. With these thoughts in mind, early reconstruction is still recommended. Functional and aesthetic outcomes should direct the extent of reconstruction to optimize the physical and psychosocial development of the child.

Many strategies have been tried for removal and reconstruction of large and giant nevi. Serial excision can often debulk these massive lesions but can rarely completely remove them. Because of their size, excision and split-thickness autografting have generally had poor functional and aesthetic outcomes. Dermabrasion, curettage, and laser treatment have problems with the reappearance of the remaining deeper nevus cells, continued risk of degeneration, and difficulty following the lesions for malignant transformation because of scarring. Even in patients in whom these surface treatments have been successful in lightening the nevus, it does not affect the tendency for some of the lesions to thicken and show heavy hair growth. The long-term effects of laser treatment on the remaining population of nevus cells must still be determined, but some experimental data suggest that it may put these cells at additional risk of degeneration.^{8,9}

We have begun staged excision and reconstruction in full-term infants as young as 6 months of age. Given the typical depth of nevocytes in congenital nevi, the excision is carried out to the level of the fascia to avoid nevus recurrence. To fill the deficit with like tissue, tissue expansion is the mainstay of reconstruction for large and giant CMNs of the head and neck, torso, and proximal limbs in our practice. We have selectively used free tissue transfer in combination with expansion to allow primary closure of donor sites or to secondarily expand free flaps in the extremities or over the neck and shoulder (shawl) areas. We favor full-thickness skin grafting over the dorsum of the hands and feet. Tissue expansion can also provide large full-thickness skin grafts with easy donor-site closure, increasing the amount of full-thickness graft available¹⁰⁻¹² (Figs. 8-4 and 8-5). In the periorbital area, we have previously reported the use of expanded supraclavicular grafts, but we now graft upper and lower eyelids separately with postauricular nonexpanded grafts. The surgical details can be found in Chapter 22.



Fig. 8-4 A, This infant with a giant CMN of the back had excision and reconstruction with nonmeshed, split-thickness skin grafts. B, The result 1 year after excision. This is one location on the trunk where excision may still yield a reasonable aesthetic outcome, whereas wrapping a graft around the flank and onto the abdomen can result in significant long-term deformity. C, The result 15 years after surgery still shows reasonably good contour and aesthetics.



Fig. 8-5 A and B, This infant had a giant nevus of the upper back, chest, flank, and anterior trunk adjacent to the breast. C, The first round of tissue expanders during expansion and just before first-stage excision. D and E, At surgery with the second round of expanders at 2½ years of age. F, Result 4 months after second-stage excision.



Fig. 8-5, cont'd G and H, A final expansion was done at age 9½ years of age to reduce scars and most of the remaining nevus lateral to the breast. I and J, Result at 11 years of age with a small area of nevus on the breast awaiting breast development before the final phase of excision.

Satellite lesions can be as large as or larger than solitary lesions, and thus excision may be warranted (Fig. 8-6; see also Fig. 8-1, B). The decision to excise satellite nevi needs to follow the previously discussed considerations, with some being excised specifically to limit the stigma that may be associated with them. Often we combine removal of the satellite lesions when we remove or reconstruct the primary lesion. Older children, who have been through previous surgeries for a large or giant nevus, are quite capable of understanding the pros and cons of additional surgery/scar and may often help make these decisions. Occasionally, changes in satellite lesions can be evaluated easily with punch biopsies rather than complete excision, avoiding some of the limitations associated with recovery from more significant surgery.

Fig. 8-6 Typical appearance of multiple satellite nevi associated with a giant nevus of the trunk. Larger satellites may be excised in early childhood in areas in which the skin may be more elastic early. Others can be followed and excised only if the child expresses concerns about the aesthetic appearance when older.



Given the prevalence of certain patterns of distribution for large and giant nevi, we have found that the planning of each stage of excision and reconstruction can be readily outlined and the expectations for the number of surgeries, expected orientation and appearance of scars in each body area, and caveats for each body region are readily outlined.

Blue Nevii

Blue nevi can be congenital but are more likely to present as smooth-domed, dark blue-black acquired nevi in the head, neck, or extremities; they occur more commonly in females. Because of the dark coloring and acquired status, concern about malignancy exists. Two histologic variants are known: the common blue nevi, which tend to be smaller and well demarcated, and the cellular blue nevi, which tend to be larger and have irregular borders and spindle-shaped melanocytes (Fig. 8-7). Both lesions are considered benign, but malignant cellular blue nevi have been reported, and thus removal is recommended.

Café-au-Lait Spots

Café-au-lait spots are well-demarcated light tan to brown macules that can be present in normal individuals or, when multiple, can be a marker of neurofibromatosis (Fig. 8-8). The texture and adnexal distribution within the macules are normal. The color may fade over a lifetime. Histologically, the keratinocytes contain macromelanosomes with increased pigment. Because of the normal quality of the skin and benign nature of these lesions, surgery is rarely recommended. Laser treatment can be considered when a lesion is present in cosmetically sensitive areas, but both recurrence and success have been reported.¹³

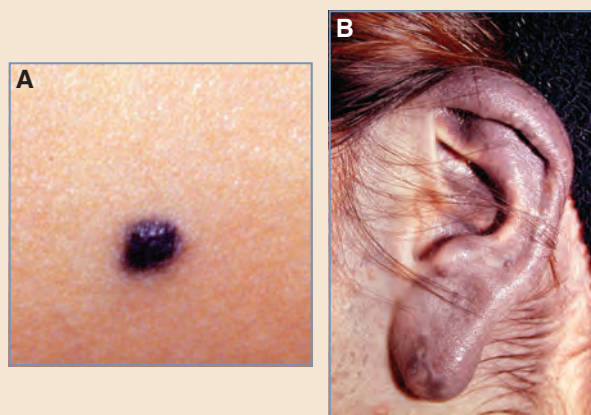


Fig. 8-7 The deep and heavy pigmentation of a blue nevus creates the characteristic blue appearance. **A**, The lesion may appear as a more typical isolated smaller lesion. **B**, It may also appear as in this rare large blue nevus involving the entire ear.

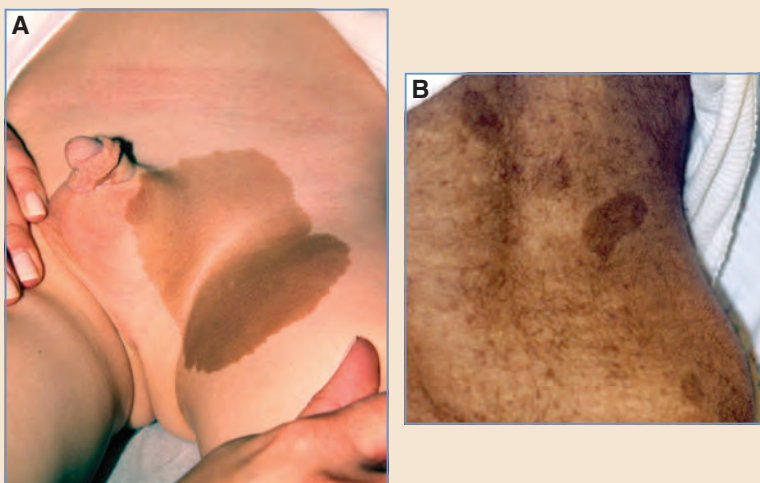


Fig. 8-8 **A**, Café-au-lait lesions may be seen as single, large, uniformly pigmented lesions on the trunk or occasionally on the face. **B**, They may also appear as multiple smaller lesions associated with neurofibromatosis type 1.

Rarely, large and giant deeply pigmented café-au-lait spots may occur in the facial area that may warrant excision. In these cases it must be made clear to the parents (or the child, if old enough) that the lesion itself carries no risk of degeneration. The benefit may come from aesthetic improvement rather than medical necessity. The family must be comfortable with the likely results of such surgery and feel that the end result is preferable to the café-au-lait macule. In these situations there is a benefit to referring these patients for initial treatment or at least testing with a laser.

Fig. 8-9 The nevus spilus, also known as a speckled lentiginous nevus, looks much like a variable area of café-au-lait pigmentation behind varied freckles, spots of pigment, or even an atypical CMN or a blue nevus. This child presented with a lightly pigmented nevus spilus with darker pigmentation. The lesion gradually darkened throughout, making the border of the lesion more distinct.



Nevus Spilus

The *nevus spilus*, also known as a *speckled lentiginous nevus*, looks much like a café-au-lait spot but has darkly pigmented areas within it, which may represent freckling, a CMN, or a blue nevus (Fig. 8-9). Suspicious areas within the nevus should undergo biopsy, and if the lesion is in a cosmetically sensitive area, complete excision can be done. Nevus spilus may on occasion be large or giant, with sizeable areas of CMNs within the boundaries. A decision may be made to selectively excise these darker areas while leaving the greater area of light pigmented lesion untreated, particularly when the larger lesion encompasses areas in which surgical scars may present later problems of a cosmetic or functional nature (for example, the perianal or genital region) (Fig. 8-10).

Mongolian Spots

Mongolian spots are blue-gray pigmented patches that present in newborn infants, particularly in Fitzpatrick type IV to VI skin types. They primarily present over the lumbosacral area but can be anywhere over the back of the body. Histologically, the melanocytes responsible for the lesion are located in the lower two thirds of the dermis. These lesions generally regress spontaneously by 3 to 4 years of age, and no surgery is indicated (Fig. 8-11).

Nevus of Ota and Nevus of Ito

Nevi of Ota and *nevi of Ito* are blue-gray to brown macules located in the distribution of the first and second trigeminal nerve branches (Ota) (Fig. 8-12) or in the shoulder area (Ito) (Fig. 8-13). All structures in the area can be involved as seen with mucosal and retinal pigmentation with nevi of Ota. They are primarily found in the Fitzpatrick IV and V skin types and are more frequent in females. Bilateral presentation can occur (10%), and concurrence with Mongolian spots has been reported. Like Mongolian spots, they represent atypical pigmentation with normal skin adnexa; however, they do not resolve over time. Although present at birth, the pigmentation may not become obvious until puberty or pregnancy, when they tend to darken. In general, these lesions are considered benign, but the development of areas of malignancy has been reported. Many modalities have been used for treatment, but laser therapy seems to be the most successful, especially with Q-switched ruby, Q-switched alexandrite, and Q-switched Nd:YAG lasers.¹⁴



Fig. 8-10 A, Nevus spilus may also present as a giant nevus with medium and large CMNs within the lightly pigmented borders. Because of both the size and marked variability of pigmentation and surface character, the managing pediatric dermatologist in this case thought that the lesion would be difficult to follow for potential changes. A decision was made to excise as much of the entire lesion as possible. B, Completion of the first tissue expansion. C, After the first excision and advancement of the expanded flaps. D, The patient is shown 2½ years after one additional expansion and excision. The remaining area of nevi is more uniform and easier to follow. Selected remaining small CMNs may be excised and closed primarily.



Fig. 8-11 Although Mongolian spots are possible in the upper back, shoulders, and extremities, they are most typically found in the lumbosacral region.



Fig. 8-12 A nevus of Ota typically presents as a blue-gray to brown macule located in the distribution of the first or second trigeminal nerve branches.



Fig. 8-13 A nevus of Ito presents as a blue-gray to brown macule located in the scapular and deltoid region.

NONMELANOTIC LESIONS

Spitz Nevus

Sophie Spitz¹⁵ originally reported this lesion in 1948 when she observed the generally benign course of lesions in children that appeared histologically similar to melanoma. Her report noted only one occurrence of metastasis in 13 cases.¹⁵ This conundrum regarding the diagnosis of Spitz tumor continues today. Clinically, the lesion typically presents as a rapidly growing, well-demarcated pink nodule with flaky skin coverage, but pigmented Spitz nevi also occur (Fig. 8-14). They can be listed in the same differential diagnosis as pyogenic granuloma. Local recurrence is a concern, and occasionally in-transit nevus cells have been noted in patients with multiple recurrences. A recent survey of practice habits of surgical dermatologists supports conservative reexcision with 2 to 3 mm margins after shave-and-punch biopsy.¹⁶

Histologically, these lesions are melanocytic nevi with specific patterns of atypia. There can be a diagnostically challenging area of crossover with melanoma that can be difficult for the pathologist to differentiate. Terms such as *Spitzoid melanoma* and *atypical Spitz nevus* should key the surgeon into the pathologist's discomfort with the diagnosis. When concern about the possibility

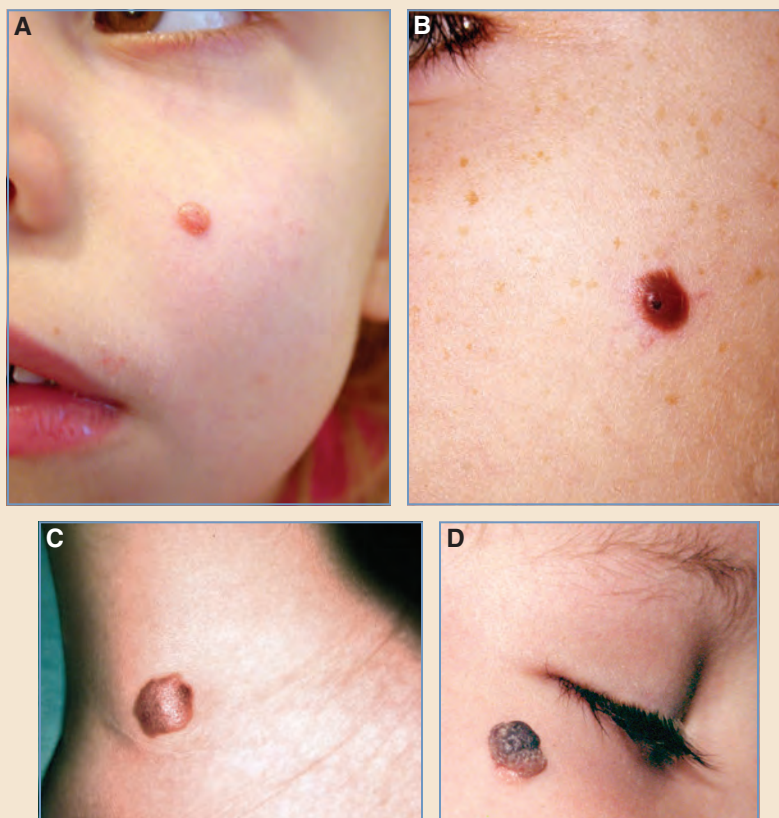


Fig. 8-14 Spitz nevi have a variable appearance. These lesions are commonly a pinkish nodule that is well demarcated and rapidly growing. **A**, This early lesion demonstrates the flaky skin surface that may also be seen. **B** and **C**, Typical pink, firm, plaque-like Spitz nevi—one on the cheek and one on the ankle. **D**, Pigmented Spitz nevi are also seen and may be confused with melanoma during visual examination.

of melanoma persists, consultation with outside dermatopathologists is useful. Sentinel lymph node biopsy has been suggested at the time of further excision in atypical Spitz nevi. However, the practice has not proved useful, and current recommendations are for clean margins and observation.¹⁷

Nevus Sebaceous

Sebaceous nevi (of Jadassohn) are seen in newborns as waxy pinkish-orange hairless plaques, usually in the scalp and face, and fall within the family of epidermal nevi (Fig. 8-15). Most lesions are isolated, but linear coup-de-saber-like lesions and extensive multiple lesions may indicate underlying ocular and central nervous system defects with associated developmental delay, seizure disorder, and sebaceous nevus syndrome (Fig. 8-16). The *cutaneous lesions* are hamartomas of malformed sebaceous units. During childhood the lesions are quiescent, but they tend to become verrucous and irritated at puberty. Reports of benign and malignant neoplasms developing within sebaceous nevi during adolescence populate the literature. Basal cell carcinoma and syringocystadenoma papilliferum are the most common neoplasms cited, with a small percentage degenerating to squamous cell carcinoma. Removal is recommended for both prophylaxis of all types of neoplasia (not necessarily cancers) and elimination of a site of irritation. The more extensive lesions also present with considerable aesthetic deformity, and treatment may at times be planned well before puberty to avoid the associated stigma that may be associated with their presence.

Although some extensive lesions have been treated with a variety of lasers, this treatment approach only temporarily manages the crusting and irritation and will need to be repeated throughout life. Surgical excision, if timed and staged appropriately, can excise even extensive lesions. In these cases we have begun serial expansion in the first year and typically combine expander placement in regions of large and giant lesions with excision and primary closure, or we stage serial excision (without expansion) for narrower linear lesions.¹⁸ Although some surgeons



Fig. 8-15 A, Sebaceous nevi are common in the scalp and facial area and vary in appearance, but typically are waxy, pinkish-orange lesions with absence of hair. B, Although some are not as elevated and distinct as seen here, the lesions often become more distinct as the individual reaches puberty.



Fig. 8-16 A and B, This infant had extensive sebaceous nevi but did not have the seizures typically associated with giant multiple lesions and sebaceous nevus syndrome. C and D, Staged excision was carried out with a combination of tissue expansion and linear partial excisions with primary closure. Here two of the expanders are in place. E-G, Approximately 6 years after first-stage surgery.

have expressed concerns about the healing of intralesional excisions, we have not found any differences in healing compared with that of pigmented nevi, if care is taken to avoid undue tension on closure. Broad-spectrum antibiotic coverage is used routinely in these cases.

Epidermal Nevi and Epidermal Nevus Syndromes

Epidermal nevi are raised, keratotic lesions that often appear in infancy and may continue to define their borders and thicken until adolescence and adulthood, making complete excision difficult in younger children. The lesions may be pigmented or flesh-toned. Like nevus sebaceous, epidermal nevi are hamartomas of skin and skin appendages; unlike nevus sebaceous, there is no predilection for neoplasia. Multiple variants exist, including the verrucous epidermal nevus (Fig. 8-17), inflammatory linear verrucous epidermal nevus (ILVEN) (Fig. 8-18), nevus comedonicus (Fig. 8-19), and Becker's nevus (Fig. 8-20). They can be isolated lesions, but when multiple or particularly large, one must be suspicious that they may be part of one of the multiorgan syndromes jointly known as *epidermal nevus syndromes*. These syndromes have ocular, neurologic, skeletal, and possible renal and cardiac anomalies associated with extensive or multiple epidermal nevi that follow the lines of Blaschko.¹⁹

Patients present for treatment of epidermal nevi because of their unsightly appearance and symptoms, such as irritation and pruritus. Multiple modalities for treating epidermal nevi have been reported, including multiple laser modalities and cryotherapy. Anecdotally, there appears to be an increased risk of hypertrophic scarring in cases of surgical excision. One specific variant for which surgery may be indicated is ILVEN (see Fig. 8-18). This is an intensely pruritic,

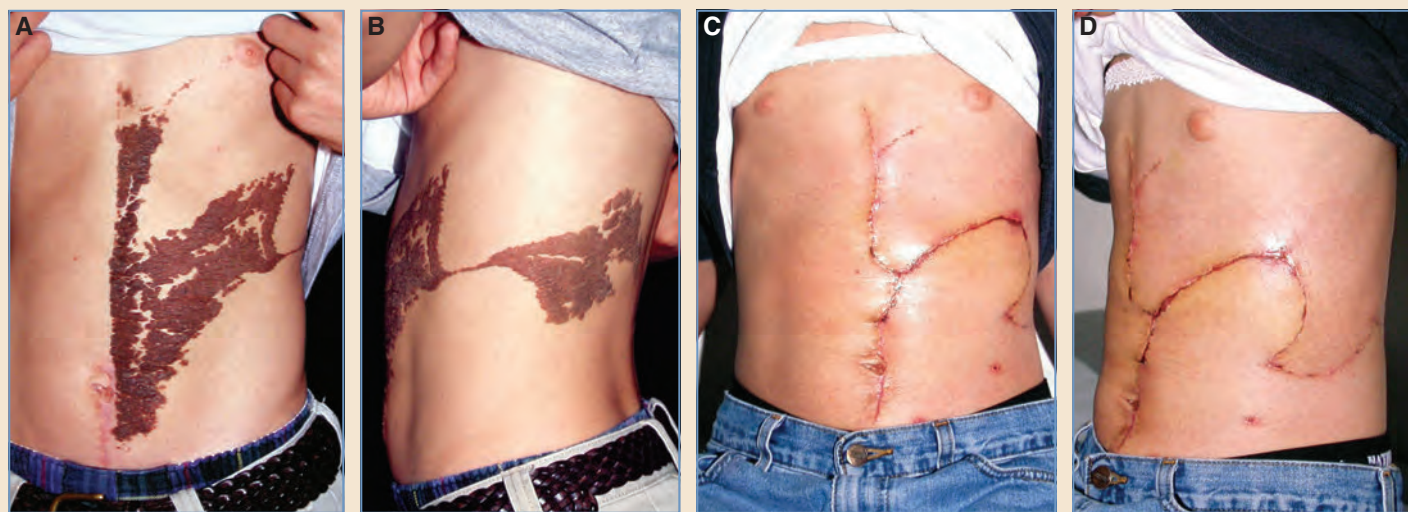


Fig. 8-17 A and B, Epidermal nevi are raised and keratotic lesions that are present during infancy and become more distinct as they thicken and become more keratotic, particularly toward adolescence. In this patient topical and laser treatments were not successful in relieving discomfort associated with the lesion, and excision was carried out, aided by tissue expansion. C and D, Immediate postoperative views after excision.



Fig. 8-18 ILVEN are commonly treated with topical medication and lasers. However, extensive lesions may be unresponsive, and patients may be “crippled” by severe pruritus and pain. **A**, As seen in this 3-year-old boy, the lesion is less distinct than the epidermal nevus in Fig. 8-17, but it may become equally plaquelike over time. Excision of ILVEN, with direct closure, skin graft, or tissue expansion, is curative. **B**, Completed tissue expansion in the adjacent thigh. **C**, Immediately after surgery. **D**, Result 1 year after excision.



Fig. 8-19 This patient has a giant area of nevus comedonicus with areas of active abscess, comedones, and pitted scar.



Fig. 8-20 Becker's nevi present most commonly as an area of pigment similar to café-au-lait spots, although with less distinct margins and heavy hair growth. These lesions are most common in the upper chest, shoulder, and upper arm.

pink, irritated lesion that weeps and crusts, is more common in females, and often is located in the lower extremity, buttocks, and perineum. There is diagnostic and treatment overlap between ILVEN and linear psoriasis. Some patients may respond or partially respond to topical agents, laser therapy, and psoriatic treatments. Failure to control the debilitating pruritus is an indication for surgical therapy, particularly those lesions in the perineal, perianal, and genital area. Patients can be totally relieved of their symptoms with excision. Unlike melanocytic lesions where small gap areas may be infiltrated with nevus cells, these segmental linear lesions can be interspersed with normal skin. Often these lesions are extensive, and planning may include excision with direct closure, skin grafting, or expanded flap reconstruction. If the full width of the lesion can be excised and closed directly or with a local flap, treatment may be accomplished in a single surgery. In other cases, several nonadjacent lesions may be excised at one procedure and remaining strips of nevus removed in a subsequent procedure. Larger lesions may require tissue expansion in the course of treatment. It is the senior author's (BSB) experience that whether young or old, these patients are some of the most grateful patients you care for, and this treatment can have far reaching benefits for the child.²⁰

Pilomatrixoma, Pilomatrixoma, or Calcifying Granuloma of Malherbe

The *pilomatrixoma* is a benign neoplasm arising from hair follicle matrix cells with a predilection to appear in areas of fine vellus hair in children. This is the most common “acquired cyst” in the pediatric population. The brow and sideburn areas of the face are particularly common sites. It presents as a hard mass fixed in the skin, often with a bluish hue or telangiectasia overlying it. Secondary infection, possibly from picking at it, is not an uncommon occurrence. These lesions grow gradually over time and rarely regress spontaneously. As they enlarge the overlying skin progressively thins, often requiring larger skin excision for uncomplicated healing (Fig. 8-21). This alone suggests some benefit of early, although certainly not urgent, treatment. Excision is recommended.²¹ When completely excised, recurrence is rare, but some patients present with second or third lesions in different areas of the face.



Fig. 8-21 A, A typical pilomatricoma, or calcifying granuloma, presents in the head and neck area as a firm cyst fixed within the skin, with a bluish-pink area of telangiectasia overlying it. In this case there is a 2 cm lobulated cyst fixed at the area of the mandibular angle showing as two points of discoloration on the overlying skin. B, A surface telangiectasia over a lesion, as in this lower medial cheek lesion, is a common development. C and D, This adolescent presented with multiple lesions that progressed rapidly to raised cystic appearances, which in some areas became inflamed, infected, and then drained.

Juvenile Xanthogranuloma

Juvenile xanthogranuloma presents as both acquired and congenital lesions in the faces of children. They initially appear as a pink nodule that becomes yellowish. Many resolve spontaneously within 1 year, but some require as much as 3 years before resolution (Fig. 8-22). If a diagnosis is made clinically, observation is warranted; however, the rapid growth of these lesions often leads to biopsy before a diagnosis is made. Histologically, they are composed of giant cells and histiocytes and are not granulomas at all. We have reported a patient in whom the orbicularis muscle was infiltrated with giant cells and histiocytes.²² Muscle infiltration has been described previously in rare truncal lesions. Rapidly growing, more infiltrative lesions may warrant early excision (Fig. 8-23). For smaller, highly visible lesions that have not resolved after 1 year, a decision may be made with the family and the child when he or she is old enough to understand the pros and cons of excision and the resulting scar to excise rather than wait for possible resolution.

Fig. 8-22 A typical juvenile xanthogranuloma presents either as a pink single nodule or multiple nodules that become yellowish, may ulcerate, and then involute. This child had just the single facial lesion.



Fig. 8-23 A and B, This infant presented with a congenital single xanthogranuloma that enlarged to involve half of her chin. During surgery it was discovered that this lesion infiltrated the underlying muscle, a phenomenon that, at the time this was reported, had only been seen with large truncal lesions. C, Appearance 3 months after excision and primary repair. D, Long-term results at 6 years of age.

KEY POINTS

- CMNs carry a small but real risk of melanoma over a lifetime. In cosmetically sensitive areas or if they cannot be easily observed, removal is recommended at an appropriate age.
- Tissue expansion can be used “primarily” to enlarge and allow transposition and translocation of local and regional flaps. Expansion can also be used “secondarily” to provide larger areas of full-thickness skin graft and to allow free tissue transfer by facilitating direct closure of the donor site. Whether used for primary or secondary purposes, tissue expansion can improve aesthetic outcomes from reconstruction of large and giant CMNs by replacing tissue with like tissue.
- Spitz nevi are benign lesions with a propensity for local recurrence. Histologically, they can be difficult to differentiate from melanomas. Review of the specimen by a qualified dermatopathologist is recommended. Sentinel node biopsy is not indicated.
- Sebaceous nevi carry a small but real risk of developing neoplasia within the lesion during the second decade of life. They also may become verrucous and quite irritated. Removal is recommended, with the timing dependent on the size and location of the nevus.
- Epidermal nevi are benign and usually can be managed with laser ablation. Before surgical resection in younger children, families should be reminded that these nevi might continue to develop. Anecdotally, hypertrophic scarring can occur after resection of these lesions, which can be problematic.
- When multiple or extensive epidermal nevi or sebaceous nevi are noted, a workup is appropriate to determine whether there is syndromic involvement of other organ systems.
- Surgical excision is not generally recommended for café-au-lait macules and Mongolian spots. The color of these lesions tends to fade, and the skin’s quality and texture are normal.
- Laser therapy is the mainstay of treatment for nevi of Ota and nevi of Ito.

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9

Acute Pediatric Burn Injury

Sandhya Deo • Joel Fish



Children are not little adults, and the differences in acute burn care for adults and children are notable. This chapter focuses on these differences and emphasizes that pediatric acute burn injury is a unique area requiring special knowledge of resuscitation, sepsis, infection control, prevention strategies, wound care, analgesia, and early rehabilitation. The long-term consequences for rehabilitation and surgical reconstruction are beyond the scope of this chapter and are discussed later in the book.

The age at which patients are no longer considered “pediatric” varies. Most agree that patients in the later stages of adolescence (14 to 16 years of age) should be considered for treatment in a pediatric versus an adult facility. Specialized pediatric burn care requires trained health personnel who can dose medications appropriately, adjust fluids, and provide necessary analgesia and sedation. Nursing and rehabilitation staff should understand developmental milestones through adolescence to ensure optimal patient care. Many adult burn care facilities have partnered with pediatric specialists to work together on the smaller population of pediatric burn patients, effectively combining their care. We think that pediatric burn patients *must* go to a pediatric facility where specialized pediatric burn care is available.

EPIDEMIOLOGY AND BURN PREVENTION

Burns are a common injury in children,^{1,2} who are naturally curious, impulsive, and active. In developed countries most injuries are small and relatively easily managed.^{3,4} However, burns are the third highest cause of accidental death in children (after motor vehicle accidents and drowning).⁵ Pediatric burns occur most commonly in the home and while children are unsupervised.^{6,7}

In the United States, approximately 500,000 burns occur annually requiring medical attention,⁸ and half are thought to be in children.⁹ In North America, pediatric burns are primarily scald injuries in the youngest age groups (younger than 5 years of age), and most occur in males;

the incidence of flame burn increases as children approach adolescence but does not reach adult numbers until they are older than 20 years of age.^{10,11}

Third world countries have a higher incidence of pediatric burns.^{6,12,13} In underdeveloped countries, malnutrition and infectious diseases, coupled with limited resources, result in high mortality rates and severe outcomes of permanent disability.^{14,15} Although the numbers are largely not known, the World Health Organization estimates the rate of child deaths from burns is more than seven times higher in low- to middle-income countries than in high-income countries.¹⁶ The lack of prevention efforts in underdeveloped countries partially explains the high rates of pediatric burn injury.¹⁷

Documenting the specific causes of pediatric burn injury is of great interest, because many injuries are preventable. The long-term impact of scarring can be significant, especially in children younger than 2 years of age, accounting for the current push to eliminate pediatric burns through education and legislation.¹⁸ Scald injuries can be reduced but are unlikely to be eliminated as long as children have access to hot beverages.

Some childhood burn injuries have been prevented because of the introduction of flame-retardant clothing, safer cooking practices, school-based fire awareness programs, education by burn prevention committees in local communities, and raised awareness of the dangers.¹⁸⁻²⁰

In North America and Europe, major pediatric burn injuries are rare; this might be explained in part by prevention efforts over the past 20 years.²¹ The decreasing incidence of major pediatric injury has resulted in difficulty maintaining competency and expertise of all burn team members to treat major pediatric burn patients. Many burn care centers struggle to ensure an adequate volume of pediatric injuries to maintain the required competency necessary for obtaining burn center verification by the standards of the American Burn Association. The use of modern telecommunication may help to link regional centers and aid lower-volume centers in maintaining pediatric burn care, preventing the need for parents and children to leave their community to receive even basic burn care.

Specialized causes of burn injury, such as chemical and electrical injury, in the pediatric population are rare in developed countries. They account for less than 1% of admissions to burn centers.²² Pediatric trauma and burn injury combined is rare.²³

ACUTE INJURY ASSESSMENT

First Aid

Basic first aid treatment applied immediately and for the correct period of time has been shown to decrease the degree of burn injury. First aid includes removing the child from the burning stimulus, removing the clothing, and cooling the burned area with cold, running water.²⁴ Caregivers might forget these simple steps in a moment of panic, and some have inadequate general first aid knowledge.²⁵ Education workshops are available for new parents. Many local burn centers that treat pediatric burn injury teach basic first aid to first responders as an effective way to reach those at risk.

The basic three steps of initial management of a pediatric burn injury are as follows:

1. The circumstance and mechanism of burn injury are assessed.
2. The burn extent is assessed.
3. The depth of the injury is assessed.

Determining the Circumstances of the Injury

Determining the circumstances of the injury in pediatric patients is imperative. Pediatric burn care providers need to understand the developmental milestones from birth to 3 years of age.

Many accidents occur at the onset of a new developmental milestone; burn injuries can occur if home care providers are caught off guard. For example, if children who are learning to pull themselves up are left alone for a few moments, they might be found upright, having reached a steaming cup of liquid and pulled it onto themselves. Knowledge of developmental milestones is essential for another reason. Therapists need to adjust their goals accordingly for children who require prolonged care over a number of weeks. Parents need to be informed that often their child will temporarily stop advancing until the acute burn injury has been treated.^{6,26,27}

Understanding the specific circumstances surrounding pediatric injury is critical in screening for neglect and abuse. Reported rates of nonaccidental injury vary among burn centers, and this is an important determinant when deciding whether to admit children with burn injuries.^{2,4,7,9} The advantage of having access to a pediatric center with a team that handles cases of suspected child abuse and neglect is that a separate team of investigative health care members can perform this function. The burn team can proceed without the circumstances of the injury affecting the provision of care. Thus the treating burn team members are not directly involved in managing the domestic situation, allowing them to maintain an ongoing, trusted relationship with the parent in a difficult situation.

Burn Depth and Extent

The relative body surface area (BSA) of a child differs from that of an adult. In children, the head is a larger proportion and the lower limbs are a smaller proportion than in adults. Methods of estimating the BSA in children include the pediatric Rule of Nines, using the palm of the child's hand as 1%, and age-specific Lund and Browder charts.^{28,29}

The basic classification of burn depth is the same for adults and children regardless of age. Accurate assessment of the extent and depth soon after the burn occurs (within the first 72 hours of injury) is frequently problematic. Most burn injuries are scalds, which can take 10 to 14 days to declare the full extent and depth of the burn. The burn depth generally is more difficult to determine in children than in adults.

The classification of burn depth based on a clinical examination within the first 72 hours of a scald injury has a low accuracy (60% to 70%).^{4,9} This leads to many issues regarding basic care models used for pediatric injuries. Burns from a scald mechanism rarely result in a perfectly uniform pattern. The pattern will typically have areas of splash marks and evidence of gravitational flow, depending on the position of the child. This can lead to an inaccurate diagnosis of the burn depth in the initial assessment in the emergency department or walk-in clinic. Superficial injuries from scald burns can be apparent for hours and may manifest as a red, painful, and blanching area. This may be completely resolved within 12 hours of the injury, adding to the unpredictable overestimation of the burned area.

The burn extent is a major factor that determines the need to triage a patient to a formal burn care center. Triage guidelines are used throughout the developed world, with minor variations in the criteria.³⁰⁻³² Most agree that a patient whose burn covers more than 20% BSA requires urgent triage to a burn care program (and typically to an intensive care–capable center for pediatric patients). Smaller injuries (10% to 20% BSA) require specialized burn care but are not as likely to require urgent intensive care monitoring. Patients whose burn covers less than 10% BSA (which describes most of the pediatric burn injuries in the developed world) require specialized burn care providers. However, they rarely need intensive care support unless they are extremely young (younger than 6 months of age) or have pediatric comorbidities that place them at risk of complications related to burn care, such as a seizure disorder, neurologic disorder, or severe developmental delay.

FLUID RESUSCITATION

The most common indication for fluid resuscitation is a burn extent beyond 20% BSA and/or full-thickness (deep) burns of more than 10% BSA. Small children younger than 1 year of age may require fluid resuscitation, including those with a burn size of less than 10% BSA, because they are particularly susceptible to the systemic consequences of an inflammatory burn injury. Crystalloid is the fluid of choice. The role of colloids in pediatric burn resuscitation is unknown, and these are not currently recommended for children with burns smaller than 20% BSA.³³

The basic pathophysiology that leads to intravascular fluid loss after a thermal injury is similar to that in adults. Fluid replacement is required. The fluid should be as isotonic as possible and should contain sodium and a buffering agent. However, children are unique in their ability to actively secrete antidiuretic hormone in response to injury; therefore they differentially retain sodium, causing water to be preferentially retained and a *hyponatremic state* to develop. This common early electrolyte abnormality is seen in pediatric patients within the first 24 hours of resuscitation. Slow correction with an intravenous saline solution helps to resolve most of the hyponatremia within 24 hours. Oral fluid restriction is never required for burns of less than 20% BSA in the absence of renal dysfunction, because hyponatremia will self-correct.

Children who weigh less than 30 kg require maintenance fluid *in addition to* resuscitation fluid, as calculated by the modified Parkland formula (4 ml/kg/% BSA).^{33,34} For children *younger than 1 year of age*, maintenance fluids (*D5 1/2 NS*) should be added according to the body weight. Dextrose is necessary to prevent hypoglycemia, because their glycogen stores are limited. A Foley catheter is inserted in all children with burns *greater than 15% BSA*, and an hourly urimeter is attached. Intravenous fluids are adjusted each hour according to the previous hour's urine output.^{33,34} In patients whose involved BSA is less than 15%, oral hydration and the continuation of breast-feeding are appropriate sources of fluid^{34,35} (although an IV line is often needed for analgesia).

The calculated ongoing requirements for fluid are often based on urine output alone for most burn patients with less than 20% BSA, with a target of 0.5 to 1.0 ml/kg/hr. Many guidelines indicate that output in pediatric patients should be 1.0 ml/kg/hr, whereas other centers routinely consider 0.5 ml/kg/hr as adequate, especially for cases with 15% BSA or less.^{33,34} Awareness of so-called fluid creep is essential. This describes the increased use of fluids because of high levels of narcotics and sedation, resulting in excessive fluid resuscitation and higher levels of abdominal and extremity compartment syndrome.³⁶ Although abdominal compartment syndrome in children has been reported, these children have extensive burns (greater than 75% BSA). Major burn injuries in which levels of fluid resuscitation approach 250 to 300 ml/kg/hr require special consideration, because these rates will not be sustained in pediatric patients.

Pediatric patients with major burn injuries require a central line, which may necessitate a specialized pediatric care unit and ultrasound guidance. Intraosseous lines can be used to temporize until definitive access can be established. Pediatric intravenous access is often a challenge in burn patients with limited available sites, where access is difficult even in experienced hands. For this reason alone, patients may be transferred to a pediatric facility where pediatric anesthetists and image-guided therapy are available.

Another nuance of treating pediatric burn injury is that, although prophylactic intubation for a burn size of more than 20% BSA is recommended for safe transport and care, often scald burns of 20% BSA can be managed in an intensive care environment with careful observation. If the fluid requirements are not high, then intubation can be avoided.³⁴ Many major pediatric burn injuries are overestimated, and the depth can often take longer than 72 hours to manifest. Flame burns in children are less common than in adults and generally result in full-thickness burn injury requiring higher levels of fluid for initial resuscitation.

PAIN CONTROL AND SEDATION

Burn injury can be intensely painful, and children especially benefit from early, adequate analgesia. This alleviates trauma and anxiety for the child and helps to facilitate ongoing treatment. Evidence indicates that multimodal therapy is most useful in pediatric burn patients.^{37,38} This includes acetaminophen, nonsteroidal antiinflammatory drugs, and the judicious use of opiates. Other medications such as methadone, gabapentin, and clonidine may be helpful but should be given only after discussion with a pediatric pain service.

Providing adequate analgesia and sedation in a controlled environment is crucial for procedures performed on the ward or in an outpatient facility, including cleansing the burn surface, debriding loose skin and blisters, and/or applying dressings. Most pediatric burn centers have their own protocol for pediatric sedation, which generally involves an opiate and a benzodiazepine.

Distraction or play therapy, for example, with toys, books, bubbles, or TV, can be used as an adjunct to decrease a child's anxiety and perception of pain.³⁸

NUTRITION

Good nutrition is imperative to optimize wound healing in all burn patients. Early oral feeding is encouraged but can be problematic in children, in part, because of the resultant gastric paresis and ileus. In this situation, nasogastric or nasojejunal tube feeding should be considered. Enteral nutrition is preferable to parenteral nutrition when possible. Early enteral nutrition preserves the mucosal integrity of the gut and improves intestinal blood flow, which prevents bacterial translocation and infection.^{39,40}

Burn injury leads to a hypermetabolic state, and a child's nutrition needs to be adjusted accordingly. The age of the child, the size of the burn, and other underlying comorbidities should be considered. Pediatric dietitians can assist in calculating the recommended daily caloric intake, with an appropriate carbohydrate/protein ratio.³⁸⁻⁴⁰

Pharmacologic agents that can reduce the hypermetabolic response include analgesics, anabolic hormones, anabolic steroids, and catecholamine antagonists. The use of insulin and/or metformin in children to stabilize blood glucose levels is well described.^{40,41} Oxandrolone, an anabolic steroid, is reported as effective for burns involving more than 50% BSA. In acute burns, oxandrolone in children has been shown to reduce hospital stay, maintain lean body mass, improve body composition, and improve hepatic protein synthesis.^{40,42-44} Supplementing vitamins and essential minerals (iron, zinc, and copper) is beneficial.⁴⁵

Children should be weighed regularly to ensure that their nutritional requirements are being met. Small children younger than 2 years of age grow at a rapid rate, and metabolic derangements have been measured for more than a year after major burn injury. Most small and moderate-sized burns do not cause the same metabolic consequences as larger burn injuries; nevertheless, supplemental feedings are needed to maintain caloric requirements.

WOUND CARE

The principles of pediatric burn management focus on promoting wound healing and easing patient discomfort. They include the following:

- Minimizing bacterial contamination
- Providing an optimal wound environment
- Ensuring adequate tissue perfusion
- Minimizing negative effects of inflammation
- Promoting reepithelialization

- Providing adequate fluid management and nutrition
- Providing adequate pain relief

After assessment, the wound or wounds are swabbed then cleaned with warm, soapy water. Debridement of any loose, devitalized skin is ideal if tolerated by the child. The wounds are then dressed. Many dressings are available. The choice varies according to the wound. In children, long-term dressings are warranted to minimize the trauma of dressing changes. Some of the available dressings are listed in Box 9-1.

The following areas in children warrant particular attention:

- The head and face
- The hands and fingers
- The feet
- The perineum
- Healed areas of skin (which require regular gentle moisturizing)

The introduction of slow-release-silver-containing products over the past 10 years has resulted in the use of a closed technique for many scald injuries. Silver products can provide sustained topical antimicrobial activity against gram-positive and gram-negative species for up to 1 week. With this treatment, many patients with burns of 15% BSA or less are managed as outpatients, as long as the pain is controlled and the dressings are used correctly. Our experience with the closed dressing technique has revolutionized care for pediatric scald injury patients, allowing early discharge, with no change in the rates of surgery or in the low rates of wound infection (Fig. 9-1). The products require proper use and need to be applied early (less than 72 hours after an injury) to be effective before the wounds are colonized. Many burn units stock one to three silver products for children. Fig. 9-2 provides a flow sheet for using the closed dressing technique. Tables

Box 9-1 Dressings Available for Pediatric Burn Patients

- Silver sulfadiazine (SSD)/Flamazine: SSD is a silver-containing cream that can be applied directly to a burn wound, with or without an outer dressing. Flamazine contains SSD and chlorhexidine for additional antimicrobial activity. The cream should be applied directly over the burn area daily. Old cream is removed before the wound is cleaned and new cream applied.⁴⁶⁻⁴⁸
- Bactigras: A Vaseline mesh gauze impregnated with chlorhexidine that provides a simple nonadherent antibacterial dressing. It requires a secondary outer dressing to hold it in place. The dressing should be changed relatively frequently (1 to 3 days).⁴⁹
- Mepitel: A porous, semitransparent mesh made from a flexible polyamide net coated with a soft silicone layer. The nonstick nature makes it ideal in children. It requires an outer dressing but can be left in place for several days if exudate is minimal.⁵⁰
- Aquacel Ag: A cellulose dressing impregnated with low-dose silver. The fibrous material absorbs exudate and allows the silver ions to be readily released. The amount of exudate will determine the frequency of dressing changes required.⁵¹
- Mepilex Ag: A nonadherent foam dressing designed to protect the wound and absorb exudate. It is composed of a hydrophilic polyurethane foam with a soft silicone inner layer and a waterproof outer layer.⁵²
- Acticoat (3 or 7 days): A nonadherent wound dressing that delivers a low concentration of silver for antisepsis. It comprises a layered mesh coated with nanocrystalline silver and an inner rayon mesh. The silver ions are activated with water.^{47,53}
- Biobrane: A semipermeable synthetic dressing made of collagen-coated nylon bonded to silicone. It facilitates fibrovascular tissue growth into the inner layer and provides a temporary bacterial barrier. It is usually applied in the operating room under sterile conditions, left intact until the wound reepithelializes. As the Biobrane lifts off, it is trimmed away. Biobrane has been shown to reduce pain and shorten healing time.⁵⁴

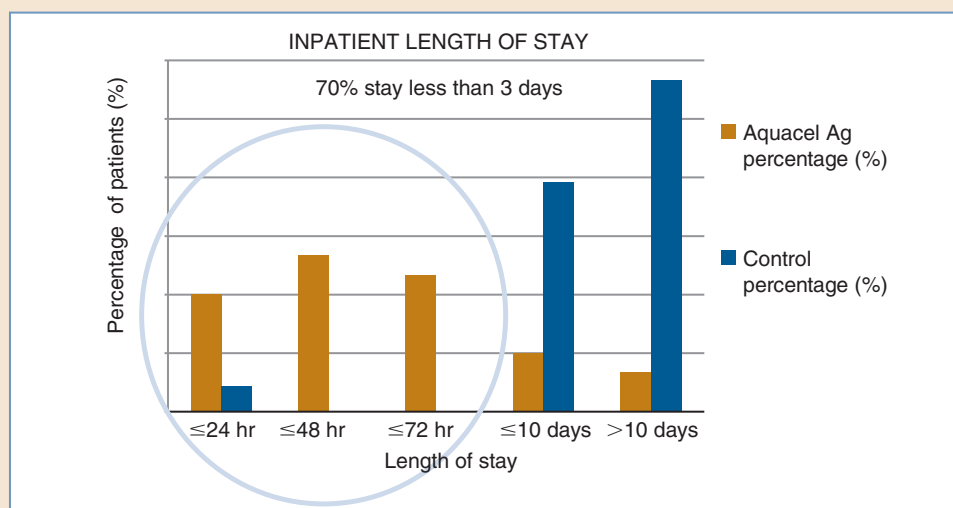


Fig. 9-1 In a historical matched-control study with 50 cases in each group, a closed-dressing technique was compared with the use of daily dressings with Polysporin or Flamazine. Results demonstrated the impact on the length of stay in a cohort of 3-year-old (average) children with an average burn size of 5% BSA. Favorable outcomes similar to these have been reported in many pediatric units.

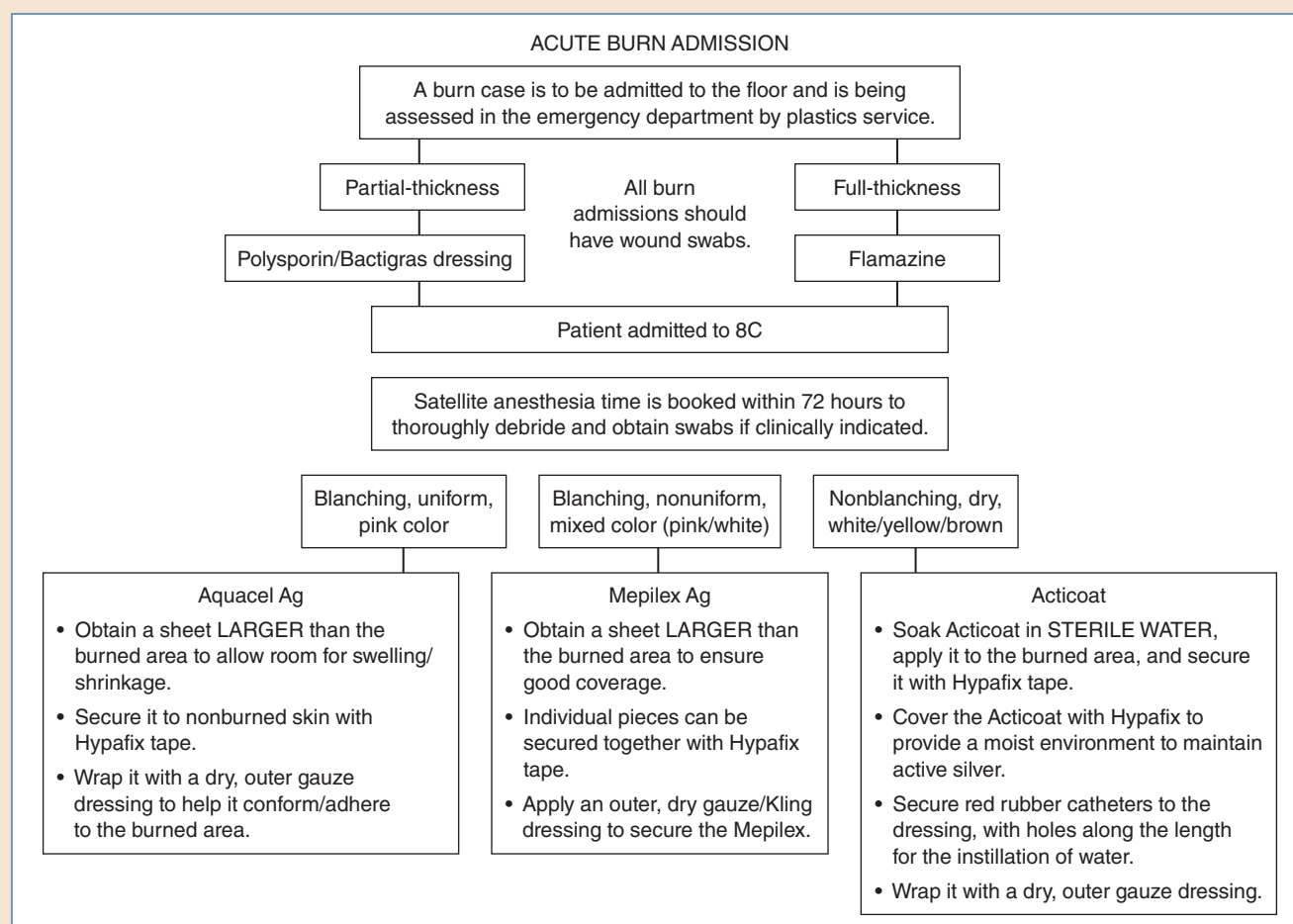


Fig. 9-2 Flow chart for using closed dressings for acute burn patients.

9-1 and 9-2 are guides that help burn care providers choose products.⁵⁵ We have provided our clinical practice guidelines, which present a rationale for product selection and use and can be adapted for any pediatric facility. Appropriate selection and use of long-acting products depends on the burn depth, which cannot be determined until 24 to 72 hours after the injury. Many cases require full sedation/anesthesia for proper application.

Table 9-1 Burn Characteristics and Predicted Healing

Depth	Color	Epithelial Budding	Capillary Refill	Healing	Scarring
Superficial	Red, erythematous	N/A	Brisk, 1-2 sec	N/A	No
Superficial partial	Pink, red	Yes	Brisk, 1-2 sec	Within 7-10 days	No, but may result in a slight color mismatch
Partial	Red, light pink, translucent white	Yes	Sluggish, >2 sec	Usually within 2-3 weeks	Possible if close to 3 weeks
Deep partial	Deep red, white, hemorrhagic staining	No	Sluggish or absent, >2 sec	Longer than 3 weeks, grafting	Yes
Full thickness	Brown, yellow, white, black eschar	No	No	Requires grafting	Yes

Modified from The Hospital for Sick Children (SickKids). Division of Plastic and Reconstructive Surgery, Clinical Practice Guideline, Toronto, Ontario, 2015.

Table 9-2 Burn Depth and Characteristics

Burn Depth	Superficial Partial	Partial	Deep Partial	Full Thickness
Appearance				
Characteristics	Red, cherry pink Blanching Sensate	Light pink, thin white eschar Blanching in areas Sensate Epithelial buds apparent	White eschar, hemorrhagic staining No blanching Sensate/insensate	Leatherlike, yellow, brown, greenish No blanching Insensate
Dressing choices	Polysporin/Bactigras (while awaiting thorough debridement) Aquacel Ag	Polysporin/Bactigras (while awaiting thorough debridement) Mepilex Ag	Mepilex Ag Acticoat	Acticoat SSD (if wound appears infected/symptomatic/presentation is delayed)

Modified from The Hospital for Sick Children (SickKids). Division of Plastic and Reconstructive Surgery, Clinical Practice Guideline, Toronto, Ontario, 2015. SSD, Silver sulfadiazine.

OUTPATIENT MANAGEMENT FOR PEDIATRIC INJURY

Most pediatric burns that occur in developed countries do not require admission to an inpatient unit and can be managed on an outpatient basis. This requires consultation and coordination between the outpatient care providers and the burn care team to ensure optimal results.

Children who do not require fluid resuscitation or supplemental nutrition should be considered for outpatient management. A suitable home situation with parents or caregivers who can support the child through the treatment is imperative. This includes the ability to attend outpatient clinics as necessary.^{56,57}

For successful outpatient management, education of the child (when appropriate) and the parents is vital. This includes how to manage the dressings between dressing changes, what to do if the dressing becomes loose, warning signs that might warrant an early return to the hospital, and the need for ongoing follow-up visits. They should be informed that care will be an ongoing process and may require long-term rehabilitation and follow-up even after the burn is healed.

INFECTION CONSIDERATIONS

Burn wounds are initially sterile but are at risk of bacterial contamination and superimposed infection. Topical antimicrobial dressings are effective in minimizing colonization and reducing the risk of true infection. Prophylactic systemic antibiotics are not warranted, and antibiotic therapy should be reserved for proven infection.^{46,47}

Infections, whether from the wound or other sources, can be difficult to diagnose in pediatric burn patients. The hypermetabolic state causes the baseline temperature to reset at a higher level and frequently leads to tachycardia and tachypnea. Continuous exposure to inflammatory mediators results in significant changes in the white blood cell counts, making these poor indicators of the presence of infection or sepsis. Other signs of infection should be used such as the appearance of the wound, cellulitis, deepening of the burn wound, increasing fluid requirements, and decreasing platelet counts.^{58,59}

If a patient has signs of sepsis, broad-spectrum antibiotics should initially be given while the cause is investigated. Burn wound infection is the most likely cause, and *Pseudomonas aeruginosa* and *Staphylococcus aureus* are the most common causative organisms. Other sources of infection are line contamination, pneumonia, and urinary tract infection. Once a source is identified and cultures have been obtained, antibiotic therapy can be specifically targeted.^{58,59}

Toxic shock syndrome (TSS) is a rare but serious infection that can develop in pediatric burn patients. It is caused by overwhelming infection by toxin-producing bacteria, usually *S. aureus*. Children are uniquely predisposed to TSS. This is thought to be the result of children not having been previously exposed to the toxin and not having developed appropriate antibodies. The mortality rate for children with TSS with a burn is approximately 50%. TSS is not limited to major burns and can occur in children with burns of less than 10% BSA.^{60,61}

ACUTE BURN SURGERY

Acute burn surgery in children has unique aspects compared with that of adults. The principles of surgery are similar, but the practices are different. Families may have difficulty understanding the difference between the burn wound and the burn scar. Discussing this concept with the family is essential, particularly with families of pediatric patients with indeterminate burns in whom the possibility of surgery is low (20%). Most families will have already explored these two entities on the Internet and have various levels of understanding. A multidisciplinary team whose members use the same terminology can help parents to understand that burn wound care and scar care are different, and that they occur at different times. Our approach has been to digitally photograph

all dressing changes and review these with the care providers so they can see the wounds and see how they have changed. This helps to focus the discussion on wound care in the primary acute stages. Spending time early in treatment to clarify this distinction is most helpful for families who suffer the loss, guilt, and traumatic aspects of their child's burn.

The timing of surgery is a major issue in treating pediatric burn injuries. The recent use of laser Doppler technology (Figs. 9-3 and 9-4) has helped to eliminate unnecessary surgery because of its improved accuracy in areas of indeterminate burn depth.^{62,63} Children are more prone to hypertrophic scar formation, and the entire process and purpose of surgery is to provide the best possible outcome of scars. Deep dermal injury increases the probability of hypertrophic scar development⁶⁴; grafting can produce a better outcome. In children, it is challenging to know how long to promote the healing phase and when to perform surgery to ensure an optimal outcome. In some cases, allowing a deep or small, full-thickness injury to heal primarily and treating the scar later is reasonable, particularly when removing the burn and placing donor skin grafts may not give a better cosmetic result. Highly functional areas such as hands are particularly problematic, because small areas of injury can result in dysfunction when not treated early enough.^{65,66}

We favor the use of sheet grafts for all cases of acute surgery until *the size of the donor site is greater than the size of the injury*; mesh grafts are then considered. All meshed grafts heal with the meshed pattern visible,⁶⁷ although those that are not spread can have an excellent cosmetic appearance. Sheet grafts require more care and expertise to apply effectively for excellent results. Each small opening in a meshed graft heals by secondary intention; therefore the scar quality is poorer than that of a sheet graft.⁶⁷ When a sheet graft vascularizes, it does not require healing by secondary intention, and less scar is formed. Sheet grafts should *always* be applied to highly visible areas such as the hands, feet, and face.

The choice of donor site in children requires careful consideration. Scalp grafts for small wounds are an excellent choice, because the donor site is completely hidden once the area heals and the hair regrows.⁶⁸ Often, adjacent skin can be selected for smaller burn injuries in children, minimizing all scarring to one anatomic area. This is useful in the short term, when the surgical dressing is confined to one area, but is more important later, when the scarring and scar therapy, if needed, are similarly limited. We do not favor the use of a buttock graft or a lateral thigh graft, which is often separate from the burn injury area. The use of the diaper region in children increases the nursing requirements for wound care and often makes ambulation and toileting difficult. For moderate-sized burns, we prefer the back as a key donor site (Fig. 9-5, A through C).

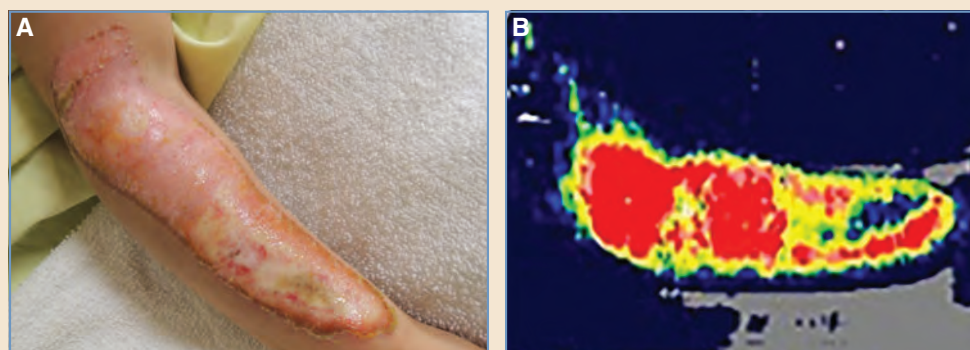


Fig. 9-3 A, This 3-year-old patient is seen 5 days after a friction burn to the forearm. B, A laser Doppler image demonstrates a lack of blood flow and a full-thickness burn in the lower forearm (*blue area*) and a high likelihood of healing to most of this wound (*red area*).

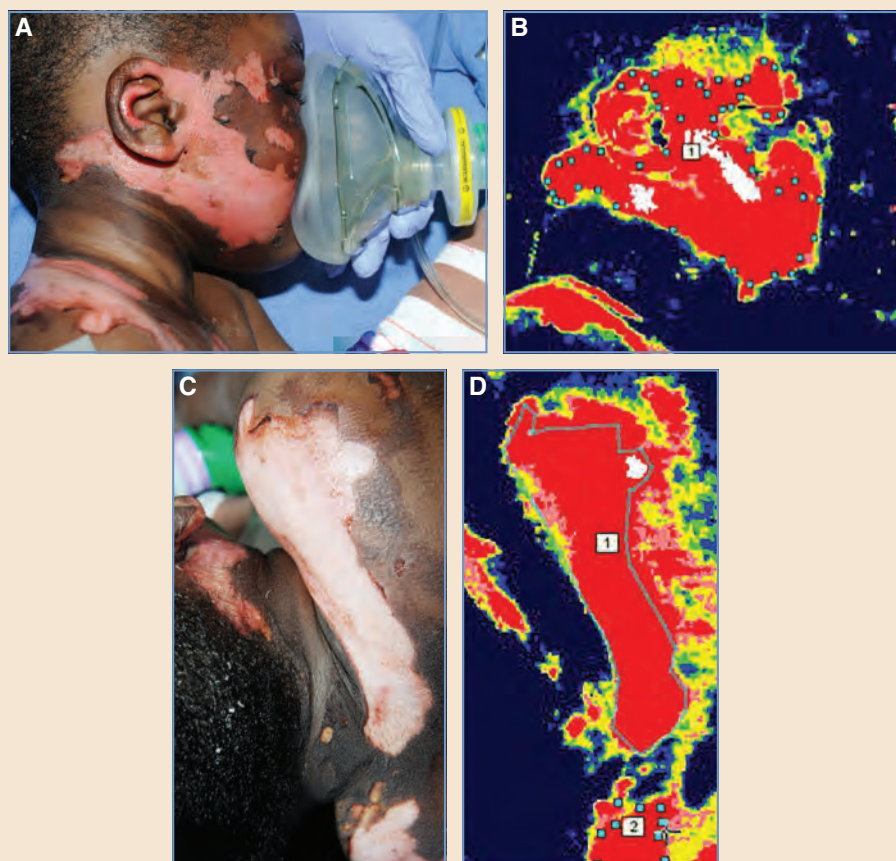


Fig. 9-4 A, This patient has a full facial scald burn and is shown 24 hours after the injury, during debridement and application of a dressing. B, A laser Doppler image indicates a high likelihood of healing to all areas, as evidenced by the *red area*. C, Second-degree burns to the shoulder area. D, A laser Doppler image of the shoulder area.



Fig. 9-5 A, This 2-year-old child had full-thickness scald injuries to the abdomen, shoulder, and arm. Sheet grafts were harvested from the back and used to cover these areas. The donor site was limited to below the scapula and out of the diaper line. B and C, He is shown 6 months after the injury. Early hypopigmentation has developed. The child is receiving laser therapy with pulsed dye and fractional CO₂ laser for severe pruritus and scar modulation.

Large, single sheets of skin are easily harvested, and the dermis, even in a child, is relatively thick, compared with other areas.⁶⁹ The patient position change to prone during the surgery does not add the same amount of time as for an adult because of the patient's small size. The principle that may need to be applied for choosing a donor site in children with a burn injury is to select, where possible, the skin that will eventually resemble most closely the area treated, which is often the adjacent regional skin.

Intraoperative hypothermia is a consideration in all burn surgeries, but young children are more susceptible than adults, particularly when large areas are burned. An operating room that has isolated and self-controlled heat to allow the environment to be warmed is an absolute necessity.⁷⁰ Underbody and overbody warmers play a role,⁷⁰ especially for patients with smaller burn injuries. However, larger burns require exposure of large areas of the body surface, and surgery is not possible unless the patients are adequately warmed. Some patients have been successfully treated with staging to minimize heat loss, especially those younger than 2 years of age. In the first stage, the lower extremities and pelvis are exposed and operated on, while the upper half is warmed. Dressings are applied. In a separate stage, surgery is performed on the upper half of the body.

The infiltration of subcutaneous tumescent solution is another essential step in safe acute pediatric burn surgery. Vasoconstriction with dilute adrenaline or phenylephrine minimizes blood loss during the donor-site harvest, and the tumescence of the tissues helps to ensure an even and consistent graft harvest.⁷¹⁻⁷³ The maximum dose of subcutaneous fluid that can be infiltrated is not known, although children are able to absorb fluid from the interstitium. However, once the fluid is infiltrated and the donor site harvested, the fluid has a natural exit point directly through the harvested skin bed; therefore the amount of infiltrate is not typically counted as part of the intravenous resuscitation. This technique requires the use of bulky absorbent dressings for the donor site and a well-adherent dressing that allows clotting when the effects of the vasoconstrictor wear off. Tumescence is a safe practice. It is inexpensive and transportable to third world countries where rates of anemia from infectious and dietary reasons are common, and blood products are not easily available. Effective topical application of an adrenaline-soaked sponge directly onto open granulation tissue before excision, without direct injection, has been reported.^{72,73}

SPECIALIZED TREATMENT AREAS

Palm Burns

Contact burns of the palm are common in young children because of their interest in exploring their environment. Most (approximately 80%) of these heal without requiring surgical intervention.^{74,75} However, burns that will require surgery need to be identified as early as possible to provide surgery and therapy in a timely fashion, with the goal of minimizing the resultant functional deficit.

Surgery on palm burns involves debridement with tourniquet control. The wound is best resurfaced with a full-thickness skin graft, which will reduce scar contraction.⁷⁶ The grafts are dressed and the hand splinted in a functional position. Where possible, the uninjured fingers should remain out of the dressing to encourage movement.

The splint is removed and the dressing debulked as soon as the graft or grafts are stable enough to tolerate movement. Young children will naturally start using their hand as comfort allows and will perform their own hand therapy; older children should begin more formal hand therapy.

Despite the best care, hypertrophic scarring and flexion contractures will develop in some cases. Aggressive scar management and hand therapy are essential. Further reconstructive surgery is commonly required as these children age and grow.⁷⁴⁻⁷⁶ The challenges of rehabilitation after surgical reconstruction for volar burns requires the specific knowledge and skill of specialized burn therapists with experience. Close observation of these children through adolescence is needed to ensure the scars do not create secondary deformity.

Breast Burns in Young Females

Burns to the anterior chest of a prepubescent female can damage the skin overlying the breast bud or the breast bud itself. Burns severe enough to require surgery should be approached in the same manner as any other skin burn. However, specific considerations are needed.

Where possible, the nipple-areola complex should be managed conservatively and complete removal of the nipple avoided. The absence of a nipple is a very noticeable deformity and can be very distressing for young girls.⁷⁷ When burns to the breast skin are debrided, the underlying breast bud should not be damaged.⁷⁷⁻⁷⁹ Any damage to the breast bud, whether iatrogenic or caused by the burn itself, will affect breast development. The burn wound should be resurfaced with a full-thickness graft where possible or a split-thickness sheet graft.

As a patient reaches puberty, the skin graft and/or burn scar might restrict breast development in this region and result in a constricted breast-type deformity.⁷⁷⁻⁷⁹ Patients should be informed that because symmetry can be affected, they may require further surgery once they have fully developed.⁷⁷ This area is a common area of injury with scald burns. The timing of reconstruction is controversial. Surgeons need to balance the needs of the adolescent female with the ability to provide a reconstruction that will last through the growth phase into early adulthood. Early referral to a breast reconstruction unit well in advance of the need for surgery is often helpful for patients who have breast scars after a thermal injury. It assists families and patients in preparing for possible future surgeries.

PEDIATRIC OUTCOMES

Patients 5 to 20 years of age have the most favorable survival outcomes from burns. Younger patients, especially infants, have higher rates of morbidity and mortality.^{5,11} As with all age groups, a larger burn extent and/or a deeper burn correlates with a poorer prognosis. Burns from house fires are among the leading causes of burn-related deaths in children. Scald burns are rarely associated with mortality.^{11,80}

There have been significant improvements in morbidity and mortality from pediatric burns over the past few decades. Advances in fluid resuscitation, wound care, infection control, nutritional support, and early surgical management (where necessary) have contributed to a significant decline in burn-related hospital admissions and deaths in children.⁸⁰⁻⁸²

The psychological impact of burns on children should not be underestimated. Children have to adjust to society's impression of their injury, especially when scars are visible. However, previous studies from the Shriners Burns Institutes show that most pediatric patients with burn injuries appear to be satisfied with their overall quality of life.⁸³⁻⁸⁶ This is especially true for children with supportive families.⁸⁷ A study published from the Netherlands in 2013 showed that although some children had long-term psychological problems such as anxiety, depression, and difficulties with social functioning, there was little evidence that behavior in general, self-esteem, or body image were impaired in the total population of children with a history of burns.⁸⁸

KEY POINTS

- Pediatric burns are common and largely preventable.
- Children who weigh less than 30 kg require maintenance fluid in addition to resuscitation fluid.
- Hyponatremia is the most common electrolyte abnormality in pediatric burns.
- Pain control is paramount for obtaining a favorable outcome.
- The closed technique for dressing changes has had a positive impact in the pediatric population.
- Use of scanning technology is helpful for assessing burn depth in scalds.
- Sheet grafts should be used wherever possible.
- Education of the child and family throughout the process will facilitate treatment and minimize the negative psychological impact.

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10

Pediatric Burn Reconstruction

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he principles of burn reconstruction in pediatric patients include (1) restoration of injured areas to as close to normal as possible for psychological and emotional rehabilitation, (2) optimization of the patient's ability to resume activities as close to preinjury level as possible, and (3) reestablishment of the natural aesthetic appearance of the child. The management of pediatric burns has changed over the years to one of early excision and coverage.¹

BURN INJURIES IN CHILDREN

Epidemiology

Of the more than 1 million major burn injuries in the United States each year, approximately half occur in children.² Although the incidence of burn injury is decreasing, these injuries still account for more than 30,000 hospital admissions and as many as 5000 pediatric deaths yearly. Burn injuries are a significant cost to society in terms of acute management, rehabilitation, and long-term care, with costs totaling \$80 billion per year.

Compared with burns in adults, flame burn injuries are associated with a relatively higher morbidity and mortality in the pediatric population. Another special issue in childhood burn injury is that although 80% of the injuries are related to accidents, up to 20% are thought to be the result of neglect or abuse.

Etiologic Factors

The most vulnerable pediatric age group susceptible to burn injury is children younger than 5 years of age. Scald, contact, and electrical injuries are most common among children. Scald injuries are more common in children younger than 4 years of age and generally involve less than 20% of the total body surface area.³ These typically result from bath water in unsupervised situations. The depth of scald injury depends on the temperature of the water, the child's skin thickness, and the duration of contact. Water at 60° C (140° F) causes a deep dermal burn in 3 seconds, whereas water at 69° C (156° F) will cause the same depth of injury in 1 second.³ Patients with scald injuries classically present with mixed burn depths. Another common scenario for scald burns is when toddlers pull containers of hot liquid from counters and tables. This produces a classic pattern of burns to one side of the scalp and face, with scalding extending down the arm to the chest.

Most massive burn injuries (that is, greater than 40% of the total body surface area) occur in school-aged children and adolescents and are commonly caused by flame burns.⁴ House fires are the most common cause of flame burns. Other sources include candles, fireworks, and campfires. The leading cause of mortality in pediatric burns is house fires, with a third of these injuries associated with cigarette smoking. Fires in enclosed areas also cause inhalation injuries, which, combined with associated direct thermal injuries, contribute significantly to morbidity and mortality. The incidence of major burn injuries has decreased over the past 10 years as a result of greater education and awareness about fire prevention.

Electrical injuries most commonly occur when children bite electrical cords or place metallic objects in an electrical outlet. The injury is usually a combination of both thermal injury from the associated flash (electrical spark) and direct injury from the current running through tissue. Older children are subject to high-voltage injuries from contact with transformers and high-tension lines.

Contact burns are fairly common, with irons and hair curlers being the most common sources. Oven doors, space heaters, and the glass fronts of gas fireplaces are frequent causes, especially for palmar burns. Hot mufflers from motorbikes, cars, and motorized equipment, especially when associated with vehicular accidents, can cause significant deep injuries.

Chemical burns can occur by contact or ingestion. The most common agents are lye-based drain cleaners, but injuries can occur from bleaches and acid cleaners.

Because an estimated 20% of pediatric burns result from abuse or neglect, physicians should suspect the possibility of abuse if hallmark patterns of injury are present or if eliciting a history from the parent or guardian is difficult. Intentional burns such as “dipping” burns, in which a child is dipped into hot water, produce a typical pattern of burns to the buttocks, perineum, and feet, with sparing of the flexion creases because of defensive posturing. Cigarette burns leave a circular, punched-out pattern. Other clues include a history of repeated trauma, blaming a child or sibling for the burn, or the burn occurring when the child is in the care of someone other than a parent.

Scarring

Unlike the process of adult scar maturation, children often undergo a more significant and lengthier inflammatory phase of wound healing. This process is followed by an exaggerated maturation phase. The entire cycle may take 12 to 18 months for the scar itself to mature. However, the fourth dimension of scar maturation is related to patient growth, which can affect scarring, because scar tissue can limit growth and produce contractures.⁵ The goal after a burn injury is a

soft, stable, pliable, avascular scar with normal pigmentation. Several preventive efforts can assist in achieving this result.

Risk Factors

The potential for hypertrophic or keloid scarring is largely unpredictable; however, there are many risk factors for abnormal scarring in a burn injury. All of the following influence the potential for abnormal scarring: the mechanism of injury (burn type), the length of time for a burn wound to heal (more than 14 to 21 days), the anatomic location (surrounding a joint, the chest, or the back), ethnicity (Asian and black patients are more likely to develop hypertrophic or keloid scarring than other ethnicities), and patient age (growing children may develop contractures, and teenage children may exhibit lengthy inflammatory scarring).⁶

Preventive Efforts

Prevention of abnormal scarring begins in the acute care setting immediately after a burn injury.⁴ Early recognition of burns that will heal spontaneously versus those that will require lengthy periods (more than 21 days) to reepithelialize is essential to minimize hypertrophic scarring. Predicting which burns will heal spontaneously often is determined by the mechanism of injury. Fortunately, most pediatric burns are scalds and should heal with meticulous wound care, good nutrition, and prevention of infection. If a scald burn does not heal by 21 days, tangential excision and grafting should be considered. Flame contact burns rarely heal spontaneously and should be rapidly excised and grafted.

MINIMALLY INVASIVE TREATMENT MODALITIES

After a burn wound is healed, either as a result of surgical excision and grafting or by spontaneous healing, the risk of abnormal scarring persists at the donor site and recipient site. In adult patients, grafting alone is usually sufficient without the need for aggressive postoperative adjuvant treatment. This is not the case in pediatric patients because of a constant dichotomy between scar hypertrophy and contraction in opposition to growing children, who may exhibit inflammatory wound healing.⁶

Pressure and Scar Massage

The use of pressure is the most reliable and proven modality for preventing or treating abnormal burn scars.⁷ This therapy can be augmented with a physical and occupational therapy program consisting of range of motion, stretching, splinting, and massage. This scar care continuum actively attempts to reduce hypertrophy and contracture and to prevent the need for reconstruction.⁸⁻¹⁰ To be effective, pressure on the scarred regions must be consistent, with garments worn for most of the day. Pressure garments must be worn in this manner until the burn scar is fully matured. Silicone gel sheets or inserts have also shown additional benefit.¹¹ Pressure garments can be customized to all areas of the body and will need to be routinely reevaluated as the child outgrows these garments. Masks should be considered for facial burn scarring (Fig. 10-1). Burns across joint surfaces should be managed with garments and splints (Fig. 10-2). Applying pressure to the wound is an integral part of care and must be reintroduced whenever a reconstruction requires a skin graft.

Scar massage with a lotion, cream, or oil adds moisture (sweat and sebaceous gland function may be poor after a burn injury) and increases blood flow to the scar, which may assist in a more rapid scar remodeling process.⁷

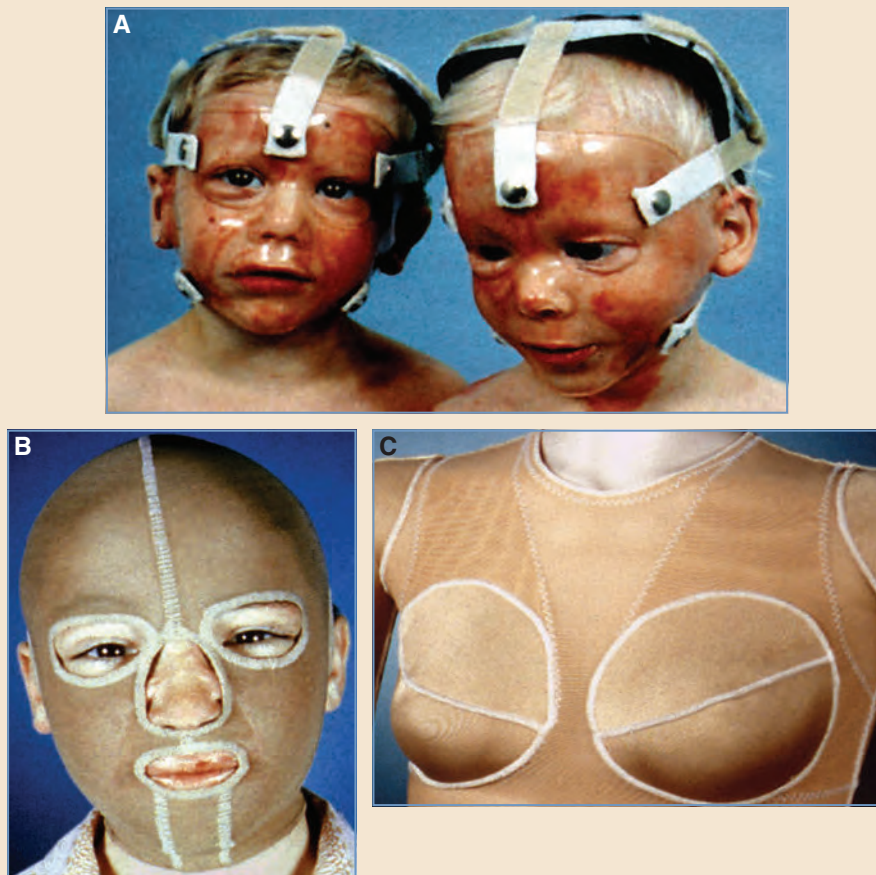


Fig. 10-1 Examples of garments and masks. **A**, Typical Uvex masks cast directly or generated by laser surface scanning. **B**, Facial Spandex mask. **C**, A Spandex bra may be used with silicone inserts. (Courtesy of Shriners Hospital for Children–Cincinnati Unit.)



Fig. 10-2 Lightweight hand splint placing the hand in a safe position.

Corticosteroid Treatment

An additional measure for managing a hypertrophic scar before beginning an aggressive surgical reconstructive program is the use of an injectable corticosteroid. For an established burn scar, triamcinolone (Kenalog, 40 mg/ml) injected intralesionally every 2 or 3 months has shown evidence of improving hypertrophic and keloid scarring.⁶ The goal of this therapy is to turn off the prolonged inflammatory phase of wound healing that increases collagen deposition and scar hypertrophy. Risks associated with this treatment are thinning of the dermis and pigmentary changes.

Laser Treatment

Persistently erythematous burn scars with or without hypertrophy may also respond to laser treatment. The 585 or 595 nm pulsed dye laser, with a target chromophore of oxyhemoglobin, has been shown to manage the persistent vascularity associated with scarring.¹² Hypertrophy associated with the scar can be reduced with the pulsed dye laser, using the laser alone or in conjunction with intralesional corticosteroid.¹³

For scar hypertrophy, stiffness, and abnormal textural change, the CO₂ laser has been used with some success. The 10,600 nm CO₂ ablative laser has been reported to improve small regions of undulation and irregularity by producing a controlled epidermal/partial-thickness dermal injury that spontaneously heals.^{12,13}

RECONSTRUCTION BY ANATOMIC AREA

Improved survival from major burns has led to increased and more complex secondary burn deformities that can significantly affect aesthetics and function. A loss or obliteration of anatomic features and structures can occur as a direct result of a burn (such as loss of an ear helix, nasal tip, or finger) or problematic scarring, resulting in restricted motion and/or growth. Despite appropriate wound care, pressure dressings, splinting and physiotherapy, troublesome burn scars can arise rapidly within the first few months after an injury.

If the tissue available for autograft is insufficient, intermediate tissue substrates can be used. Integra is an example of a bilayered, collagen-based dermal regeneration template. In children it is used with favorable outcomes in acute burn care and burn scar revisions.¹⁴ Integra is just one of many different kinds of dermal regeneration templates available.

Growth, development, and inconsistent patient cooperation should be routinely considered when planning the reconstructive needs of a burned child. Reconstruction generally takes place after scar maturation is complete. Two areas may require earlier intervention in burned children: the neck and eyelids.³ Because of significant restriction in range of motion, neck contractures can complicate general anesthesia by causing airway access issues; thus they require prompt attention. Contractures of the eyelids can cause exposure keratitis and corneal ulceration. Timely reconstruction is needed to prevent loss of vision.

Head and Neck

Cosmetic deformities in the face are generally combined with functional concerns, given the importance of facial expression and appearance in everyday social interactions. Furthermore, it is challenging to camouflage burn scars on the head and neck with clothing or coverings. The components of the head and neck that are to be considered when planning a reconstruction are the face (eyelids, eyebrows, mouth, ears, nose, and jaws), scalp, and neck. Definitive reconstruction of the burned head and neck should be postponed until skeletal maturity.



Fig. 10-3 Release and resurfacing of the lower lip with an expanded neck flap. **A**, Preoperative extensive scarring of the chin. **B** and **C**, Postoperative resurfacing with expanded neck skin. This patient also required a bilateral sagittally split mandibular advancement.

Face

The approach to resurfacing should follow the unit and subunit approach of facial aesthetics first described by González-Ulloa in 1956.¹⁵ The goals of treatment are the following¹⁶:

- A face that is normal at a conversational distance
- Maintained facial balance and symmetry
- Distinct aesthetic units fused by inconspicuous scars
- A doughy skin texture appropriate for corrective makeup
- Dynamic facial expression

Consideration of the location and direction of the borders of skin grafts is essential to ensure that scars are concealed along natural skin tension lines and anatomic borders. The areas most likely to cause secondary functional deficit are the skin surrounding the eyes, nose, lips, and mouth rather than the cheeks or large, flat areas. Appropriately placed Z-plasties can prevent scar band formation between the aesthetic facial subunits. To minimize scarring, mesh grafts should not be used in the face, and full-thickness grafts should be used whenever possible.

Sheet grafts, either full- or split-thickness, are preferred for resurfacing. Given the contours of the face and difficulties using bolster dressings, vigilant daily aspiration of any collections under the grafts is required. If the neck is spared, the surgeon can expand the neck skin to advance to the face (Fig. 10-3), but this may cause additional problems by placing tension on the mouth and eyelids. Large cervicofacial flaps can also be used.

Eyelids

Eyelid contractures, primarily of the lower lid, can lead to ectropion and corneal exposure.¹⁷ Although tarsorrhaphy can be a temporary solution, its use should be limited to the acute care

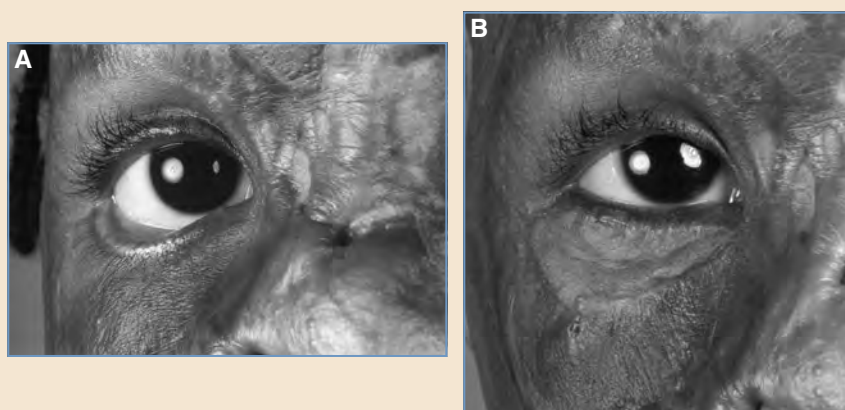


Fig. 10-4 Lower eyelid release with a graft. **A**, Preoperative lid retraction. **B**, Postoperative result.

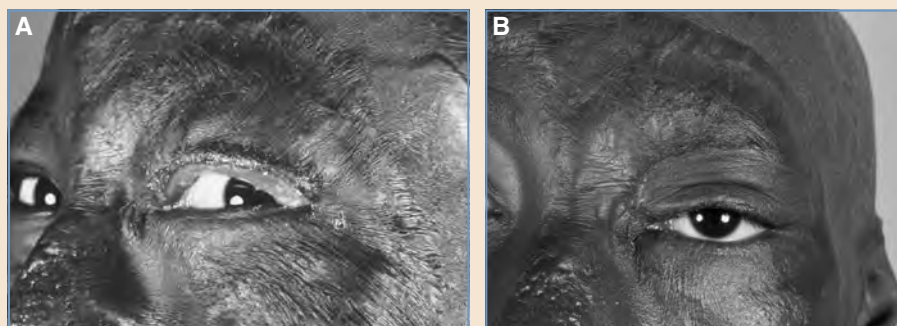


Fig. 10-5 Upper lid contracture. **A**, Preoperative contracture. **B**, Postoperative result.

phase, and the lid should be reconstructed or resurfaced as soon as possible.¹⁸ Ectropion may be either intrinsic or extrinsic. With intrinsic contracture, the ectropion is caused by scarring and shortening of the lower lid itself. In extrinsic contracture, scarring of local tissues in the cheek or neck is responsible, despite an uninjured lower eyelid. Extrinsic contractures can also occur from reconstructive efforts, such as local tissue transposition or expansion that leads to unintentional pull on the lower lid. The initial treatment should be release of the primary site of contracture, whether it is the lid itself or a distant site. This usually corrects the ectropion. If adequate lid release is not obtained by releasing the extrinsic contracture, full-thickness grafting of the eyelid is frequently necessary.

If upper eyelid skin is inadequate or unavailable because tissue has been burned, other favorable donor sites include retroauricular skin or the inner upper arm (Fig. 10-4). Occasionally, a laterally based unipedicled Tripiier flap with a canthopexy will elevate the lateral canthus and release and reposition the lower lid. If middle lamellar damage occurs, support for the lower eyelid can be improved with conchal cartilage or deepithelialized hard palate mucosa.

Upper lid retraction and resultant lagophthalmos is a less common cause of corneal exposure. Treatment is similar to that of ectropion, involving release of the contracture with inlay grafts (Fig. 10-5). If both eyelids are involved, the best results are obtained by staging the releases.

Entropion can also occur from a burn injury, but it is less common than ectropion. The pathophysiology differs from that of the typical congenital involutional or cicatricial forms seen in nonburned individuals. Patients with entropion typically have a normal underlying lamella, but the lid is rolled in by transverse shortening caused by scar contracture. This is corrected by releasing the transverse bands, usually with grafting. This allows the lid to lengthen and redrape. If this fails and the lid is of normal transverse length, a typical tarsal procedure may also be necessary.

The total loss of eyelids is fortunately rare, but if it occurs, the destruction of surrounding structures usually precludes local flap reconstruction, and a free tissue flap must be considered.¹⁹ The surgeon should take into account drainage, lubrication, and an opening for sight.

Eyebrows

Reconstruction of the eyebrows depends on the extent of the loss, the condition of the underlying tissues, and the wishes of the patient.^{20,21} Tattooing can offer aesthetic improvement, particularly in less favorable scar beds. If the bed is adequate with good vascularity, micrografts can be attempted. For more extensive loss, free strip grafts harvested from the occipital or parietal scalp can be used (Fig. 10-6). These grafts should be less than 1 cm wide, because vascular ingrowth occurs initially from the recipient site margins, not the base. Care should be taken to orient the strip graft to mimic the normal direction of hair growth. In the most severe cases with loss of supporting tissue, reconstruction can be accomplished with pedicled vascularized strip grafts based on the temporal artery²² (Fig. 10-7).

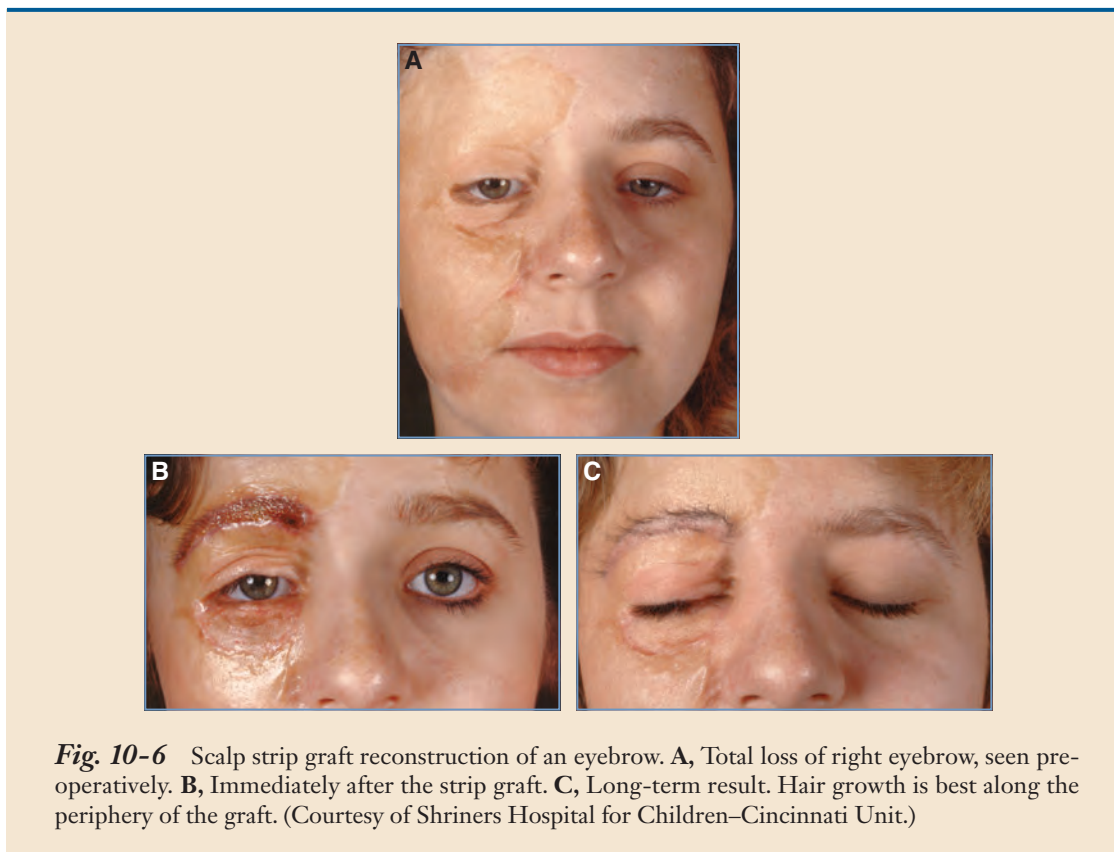


Fig. 10-6 Scalp strip graft reconstruction of an eyebrow. **A**, Total loss of right eyebrow, seen pre-operatively. **B**, Immediately after the strip graft. **C**, Long-term result. Hair growth is best along the periphery of the graft. (Courtesy of Shriners Hospital for Children–Cincinnati Unit.)



Fig. 10-7 Pedicled hair-bearing temporal artery flap for eyebrow reconstruction. **A**, Preoperative defect. **B**, Immediately after inset. **C**, Final result. (Courtesy of Shriners Hospital for Children–Cincinnati Unit.)



Fig. 10-8 Staged unit release of the upper and lower lip. **A**, Preoperative lower lip ectropion. **B**, Following unit release of the lower lip. **C**, The lip healed before the upper lip unit release was performed. **D**, Following upper lip release. **E**, Long-term result.

Mouth

Reconstruction of the mouth should be separated into the lower and upper lip if reconstruction of both is required (Fig. 10-8). A common problem in the upper lip is thinning or loss of the philtral dimple (Fig. 10-9), which is difficult if not impossible to correct. In the lower lip the pull of the scar often dramatically lengthens the lip transversely. It is often helpful to shorten this distance with a midline vertical wedge resection at the time of unit release. This allows the lip to rise and restores oral competence. Tight commissures will usually persist after release of the lips and will have to be addressed separately. They can be released with either separate inlay skin grafts



Fig. 10-9 Darting a unit release of the upper lip. **A**, Preoperative retraction of the lip and nose. **B**, Immediately following release (with extension across the nasolabial folds). **C**, Long-term result. The lip is elongated and thin, with loss of the philtral dimple.



Fig. 10-10 Release of perioral banding with Z-plasties. **A**, Preoperative markings of planned Z-plasties. **B** and **C**, Postoperative results.

or mucosal advancement flaps. Perioral banding from circumferential oral burns causes special problems by restricting the opening, restricting chin and mandibular growth, creating lip ectropion, and creating malocclusion from pressure on the teeth (Fig. 10-10; see Fig. 10-9). Commisuroplasty can be performed to restore the features of a normal mouth with a musculomucosal advancement flap in Y-to-V fashion.

Ears

Reconstruction of a burned ear is challenging because of scarring that prevents facile redraping of local skin over new cartilage frameworks. Burns commonly destroy the helix and leave the conchal bowl intact. The thickened surrounding skin may be delayed and rolled to create an acceptable helix. For more extensive loss, a typical cartilage framework can be carved from rib and covered with a temporoparietal flap and split-thickness skin graft (Fig. 10-11). In the most severe burns, prosthetic ears can be used with bone anchors if needed.²³

Nose

Nasal reconstruction is one of the most important elements in rehabilitation for patients with devastating deformities of full-thickness facial burns. The burned nose is a prominent feature of the face that presents reconstructive challenges, including total or partial loss, obliteration of features, and stricture of the airway. When possible, we use a single pedicle for complete nasal

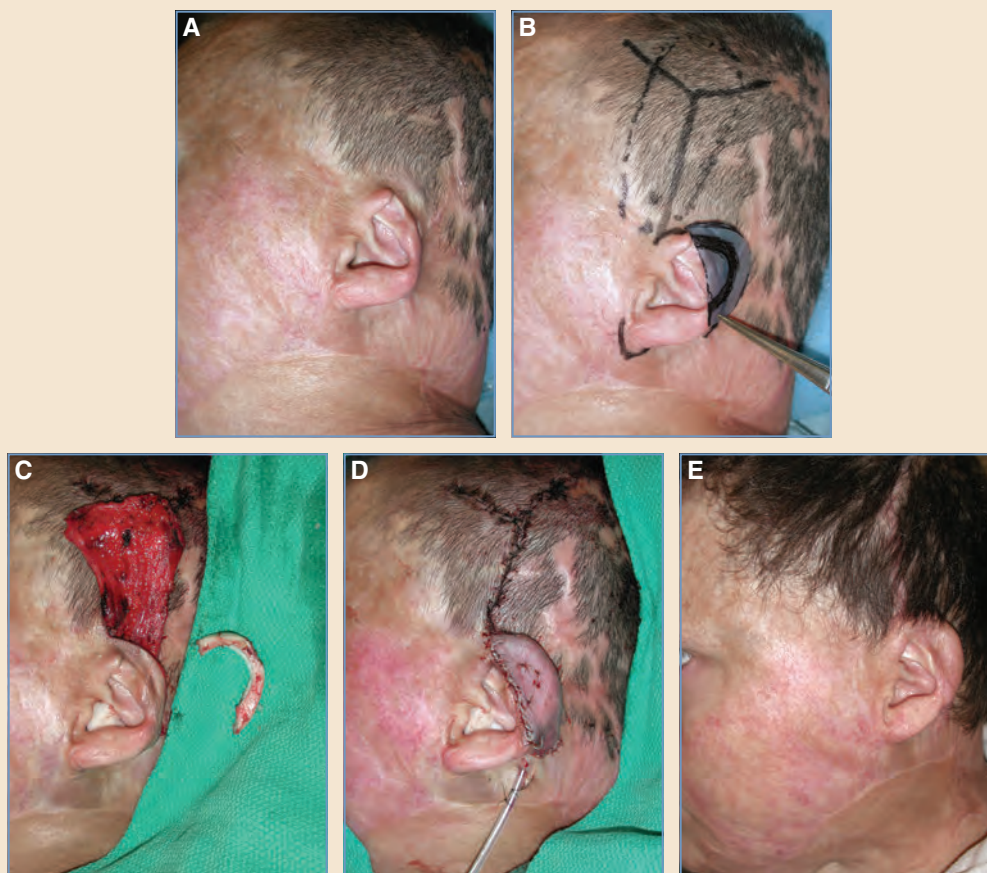


Fig. 10-11 Reconstruction of ear with temporoparietal flap. **A**, Burned ear defect. **B**, Raising the temporoparietal flap. **C**, Rib cartilage graft. **D**, Rib graft covered with a flap and a split-thickness skin graft. **E**, Final result.

reconstruction. The forehead generally furnishes the best donor area. More distant donor sites are used if the forehead is not available. In lesser deformities, resurfacing with composite grafts or full-thickness skin grafts gives satisfactory results. Nasal reconstruction in children should be staged, with the nasal dorsum contracture addressed in the first stage and the final correction performed in the second stage once the face is mature. Scars and grafts in the area of the nasal dorsum and glabella produce contractures that result in flattening and widening of the nasofrontal angle. Modified Z-plasties can be used to correct the nasofrontal deformity.

Nasal burns also cause retraction of the nares and obliteration of the alar bases. Frequently, this can be improved with simple unit resurfacing, turning the margins of the nares down to correct the retraction. Obliteration of the alar bases is very common and difficult to correct. Improvements can be made by releasing the base and placing a small full-thickness graft below the alar base to re-create the natural curve. For total or partial loss of the nasal structures, surgeons must borrow from experience with skin cancer patients. Lining, cartilage support, and covering must be supplied. Ideally, the forehead is the perfect donor site, but with burns severe enough to result in nasal loss, the forehead is rarely spared.²⁴ If it is available, however, it should be used. Free flaps, typically radial forearm, can be used for total nasal reconstruction in conjunction with

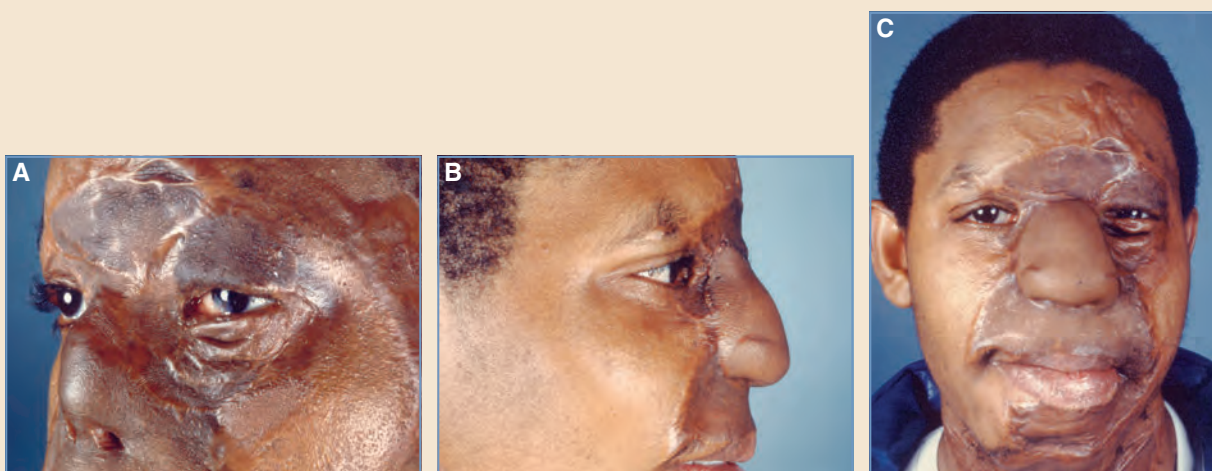


Fig. 10-12 Total nasal loss reconstructed with a free radial forearm flap. **A**, Initial defect. **B** and **C**, Postoperative reconstruction with turn-down flaps for lining, rib grafts for skeletal support, and a free radial forearm flap for coverage. (Courtesy of Shriners Hospital for Children–Cincinnati Unit.)

rib grafts to provide both bony and cartilaginous support (Fig. 10-12). Limitations of donor sites may also necessitate the use of a classic Tagliacozzi nasal reconstruction or distant tubed pedicled flaps brought up on wrist carriers.

Nasal stricture caused by collapse and hypertrophic scarring can be released and treated with full-thickness inlay grafts followed by prolonged splinting.²⁵ If an unburned ear is available, a composite chondrocutaneous graft harvested from the anterior surface of the triangular fossa provides a useful inlay graft with minimal subsequent contraction.

Jaws

The forces generated by burn scars and the associated treatment, including pressure garments and masks, often can have a profound effect on growth and development of the maxillofacial skeleton. The forces of scars on the facial skeleton can lead to malocclusion and retrusion of the mandible. Correction involves release of the deforming force, whether it is a scar or a mask.²⁴ Typically in the lower face, pressure from a burn scar or mask will initially result in loss of chin prominence. If the force is persistent and sufficient, it will eventually change the occlusion to Angle class II. Constriction from facial burns can also restrict growth and alter the direction of maxillary growth, resulting in class III occlusion and/or open bites (Fig. 10-13). In all cases, the restricting forces should be released and orthognathic surgery performed as necessary to correct the deformity. Overcorrection with osseous genioplasty is an established approach for managing a postburn retrusive chin.²⁶

Scalp

Until the introduction of tissue expansion, scalp reconstruction was limited to staged excision of alopecia, a Juri flap^{27,28} transposed to create a hairline, and Orticochea flaps to redistribute the remaining hair in a single stage.²⁹ The unique nature of the hair-bearing scalp precluded distant flap or graft reconstruction. Tissue expansion has basically eliminated the need for most local flap procedures.³⁰ Tissue expansion is well tolerated, can be repeated multiple times, and replaces like with like tissue. Serial expansions can be used to reconstruct losses of up to 50%



Fig. 10-13 A difficult case of an adolescent male who fell into a cooking fire as a young child. His chin was scarred to his chest and resulted in elongation of his mandible, retraction of his maxilla, a 34 mm occlusal underjet, and severe neck and facial scarring. **A** and **B**, Following extensive neck release. **C**, An intraoperative bilateral modified Weber-Ferguson approach to the maxilla was necessary because of severe scarring and contracture. **D-E**, Soft tissue release and resurfacing were necessary after a LeFort I advancement of 17 mm and a sagittal split mandibular setback of 17 mm. **G** and **H**, Final result. (Courtesy of Shriners Hospital for Children–Cincinnati Unit.)

or more of the hair-bearing scalp. Factors to consider are the quality of the remaining scalp, the hair density, and the state of the burned scalp. Expansion does not create new hair follicles; it only spreads out remaining follicles. For example, thin blond hair may appear sparse after several expansions. Spotty burns can be difficult to reconstruct, making large expansions difficult. The condition of the burned scalp is also a major factor. Skin-grafted, partial-thickness loss and a grafted galea are usually stable. Skin-grafted periosteum or grafts on decorticated bone can be unstable and prone to breakdown. Although expanded scalp is ideal for covering these unstable areas, the lack of suitable donor scalp may require free flap reconstruction such as a latissimus dorsi flap with skin grafting.

Tissue expansion involves placing one or more expanders around the area of alopecia (Fig. 10-14). Ideally when planning, surgeons should take into account the available unburned scalp, the direction of the hair growth, the possibility of future male pattern baldness, and the density of hair growth. In severe loss, this may not be possible. In general, the largest possible expander should be placed adjacent to the area of alopecia. Textured expanders and expanders with backing should not be used. The complication rate in children is high; complications include infection, exposure, and leaking.^{31,32} To reduce complications it is helpful to let the wound heal fully (about 3 weeks) before beginning expansion. The incision for the insertion should also be carefully planned. It should be in healthy, thick, well-vascularized tissue. Typically it is placed along the margin between the burned and unburned scalp. Scalps can be reexpanded multiple times.³³ If the first expansion does not provide sufficient coverage, a tissue expander can be reinserted at the time of removal and the first attempted advancement. This, however, carries a much higher complication rate, and most surgeons prefer to allow the wound to heal and stabilize before starting another expansion of the same area.

Trunk and Breasts

Burns to the trunk in children are generally from ascending flame burns from clothing or descending hot fluid pulled from above. Boys and girls have negative cosmetic effects from disfigurement of the trunk. Scar band contracture of the trunk is usually less of an issue than when it occurs on extremities, because the range of motion is greater with extremities. Symptomatic bands typically occur along the flanks and across the back between the shoulders. The bands often tighten during growth spurts, although mere tightness is not always an indication for release. The bands may stretch, but they do so at a slower rate than unburned skin. Surgeons should wait about 6 months to determine whether they relax on their own. The bands are released in standard fashion with inlay grafts or local flaps.

Reconstructive surgeons must consider an additional psychological stressor in girls: the loss of femininity for the developing female with a deformed breast. During the acute treatment phase, excision of severely burned tissue is necessary. Aggressive debridement is associated with a higher incidence of loss of breast tissue. Allowing the eschar to fully demarcate before tangential excision optimizes the debridement. Many surgeons suggest that debridement should be done conservatively or not at all within 2 to 3 cm of the areola, because the entire breast will arise from the breast bud beneath the nipple-areola complex (NAC).

After the initial debridement and coverage with split-thickness skin grafts, patients will either develop a normal breast that may be hindered by scar contracture or have deficient breast development from extensive debridement or burn injury. The reconstructive pathway is usually initiated with release of scar tissue in the area of the breast. Most patients undergo surgery during the younger teenage years.³⁴ Release surgery can be performed when breast tissue bulges through unburned areas, but timing is greatly based on the surgeon's best judgment.



Fig 10-14 A, This 9-year-old boy had a flame contact burn as a toddler that developed preauricular and postauricular hypertrophic scarring. B, Placement of a cheek tissue expander (filled midway). C and D, The expander was completely filled. E and F, The tissue expander was removed, the burn scar excised, and the flap advanced. G and H, He is shown after reconstruction.



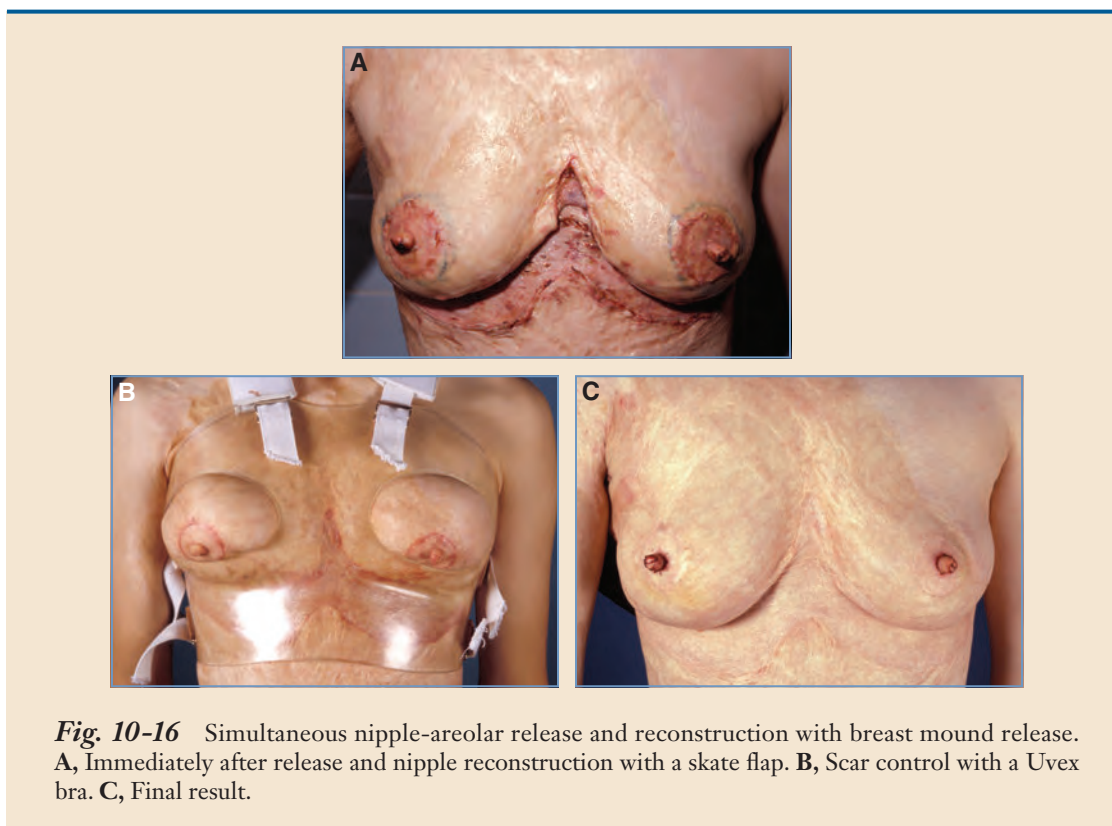
Fig. 10-15 An unusual case of bilateral burned breasts complicated by tubular breast deformity. **A**, Preoperative burn and congenital deformities. **B**, Following extensive release and resurfacing of the breasts, showing the extent of the congenital deformity. **C** and **D**, Following secondary correction of the congenital tubular deformity.

The locations of scar releases are typically in the inframammary crease, between the breasts, and around the areola. Postoperatively the grafts are placed in compression garments with silicone inserts to apply appropriate pressure to the graft sites. Ideally, the constricting envelope is released when the breast is fully developed (Fig. 10-15). In severe situations release may be required early and may have to be repeated. Release can also be combined with resurfacing if needed and if skin is available.

Most female patients with burn injury to the breasts will undergo the inframammary releasing procedure in their early teens. However, for those requiring greater reconstructive needs, it is wise to wait until the patient is in her early twenties. This allows full, natural breast development. Once full breast development is complete, several options are available for augmenting the breast tissue that is present.

Breast reconstruction options include autologous and implant-based reconstruction or a combination of the two. The size of the remaining breast, the elasticity of the chest wall and overlying skin, and donor site availability must be evaluated. The skin is often thin and friable much like irradiated skin. Therefore the risk of expander erosion, skin necrosis, and infection is higher than in normal skin. If the grafted skin is of good quality and texture, expanders should be placed in the submuscular position.

A flap is usually necessary in situations in which the skin graft has been placed on the ribs with minimal pectoralis muscle present. The most commonly used flaps are the latissimus dorsi flap and the transverse rectus abdominis myocutaneous (TRAM) flap. The latissimus dorsi flap provides a natural breast mound that is a reliable bed for NAC reconstruction. Although the TRAM



flap is a viable option for postmastectomy patients, it may pose challenges in pediatric patients, because they have insufficient abdominal laxity (especially if nulliparous), and they may not have enough tissue for viable breast reconstruction. Furthermore, patients with burns to the breasts often also have burns in the abdominal area, limiting the use of the TRAM flap.

Regardless of the reconstructive approach, patients will require NAC reconstruction of the affected breast and probably a balancing procedure if the contralateral breast is unaffected. The NAC can be reconstructed with the inframammary release if the nipple-areolar position is easily ascertained, but in some cases a periareolar release is also necessary (Fig. 10-16). Any remnant of the patient's own nipple or areola should be used and reconstructed. In general, with a total loss of the NAC, the reconstructed complex should be placed in the ideal aesthetic site once the breast mound is fully released. Because of scar contracture, burned breasts are less likely to develop ptosis. If a patient can feel a nipple sensation when the nipple is not apparent on gross examination, the nipple-areolar reconstruction should be centered around this site. Nipple-areolar reconstruction techniques are similar to those used in breast cancer reconstruction. Grafts and tattooing are used for areolar reconstruction, and local flaps are used for nipple reconstruction (bell, modified star, and skate flaps). Skate and star techniques are reported to have the best long-term projection.³⁵

Future pregnancy is an additional concern for young girls with burns to the trunk. However, the constricting abdominal scar is not associated with an increased risk of complication, compared with the unburned population. Two large series, one from the Houston Shriners Unit³⁶ and one from the Cincinnati Shriners Unit,³⁷ investigated this issue. Both series studied the effects of large burns (greater than 50% of the total body surface area) and circumferential burns on childbearing. Despite differential expansion of the abdomen caused by the presence of burned

and unburned areas, the patients mirrored the unburned population in complications, the incidence of vaginal versus C-section delivery, and the ability to carry. There have been scattered reports of a need for surgical release of the expanding abdomen and postpartum release, taking advantage of the stretched unburned skin. Pregnancy in healed burned patients usually does not require any subsequent intervention.

Hands

As with facial burns, reconstruction of hand burns starts during the acute care period. Proper management at this stage reduces the need for later reconstruction and enhances functionality. Hand burn injuries should be kept elevated above the heart, rapidly debrided, and resurfaced. The hand should be placed in a safe position. Although pinning joints may be necessary, pins should be removed early, and an aggressive splinting and exercise program should be instituted. Hands should be resurfaced with sheet grafts wherever possible, especially if they do not heal within 14 days. The close proximity of vital structures (that is, joints and tendons) to the surface of the skin increases the likelihood of damage to these structures and a subsequent need for acute or early flap reconstruction. Early mobilization of the joints in a burned hand ensures the best chances for restoration of function.

McCauley³⁸ has studied and classified the correlation between the extent of burn injury to the hand and functional outcome. Category I injuries required no surgical intervention in the management of superficial partial-thickness burns; almost all (97%) of these patients had complete functional return. Patients with category II injuries required skin grafts for closure; most of these patients (81%) were able to achieve nearly complete functional recovery. Patients with the most severe injuries involving tendons, joints, or bone were assigned to category III; very few of these patients (9%) were noted to have returned to normal function.

The multiple joints and web spaces of the hand are prone to banding and contracture. With regard to reconstruction, three categories should be considered in the hand: soft tissue deformities, joint deformities, and amputations. Contractures of the hand can be classified using the McCauley classification in Table 10-1.³⁸ Release of these contractures follows the standard tech-

Table 10-1 McCauley Classification of Burn Scar Contractures in the Hand

Grade	Definition
I	Symptomatic tightness but no limitation in range of motion Normal architecture
II	Mild decrease in range of motion without significant impact on activities of daily living No distortion of architecture
III	Functional deficit noted, with early changes in the normal architecture of hand
	A. Flexion contractures
	B. Extension contractures
	C. Combination flexion and extension contractures
IV	Loss of hand function with significant distraction of normal architecture of the hand
	A. Flexion contractures
	B. Extension contractures
	C. Combination flexion and extension contractures



Fig. 10-17 An unusual case of a palmar-burn claw hand. **A** and **B**, Preoperative extent of the deformity. **C-E**, Postoperative views after release using multiple sheet grafts.

niques of Z-plasties, local flaps, and grafts^{39,40} (Figs. 10-17 and 10-18). Because the dorsum of the hand is more likely to be injured, banding typically occurs at the juncture of the burned dorsum and the unburned palmar surface. This is an ideal situation for using Z-plasties, taking advantage of the unburned palmar skin for one limb of the Z-plasty. The Zs are typically placed at the level of the joint crease. The resulting middle limb blends with the naturally occurring crease. Because of its broad expanse, the first web space responds well to either a four-flap Z-plasty or a double-opposing Z-plasty (Fig. 10-19). The other webs are narrower and deeper and usually require a local rotation flap (Fig. 10-20), goalpost flap, or skin graft release (Fig. 10-21), depending on the extent of the contracture and the quality of the surrounding skin.^{41,42}

Dorsal hand burns tend to be more severe than palmar burns, because the skin is thinner and more exposed to injury. The resulting contractures involve hyperextension of the wrist and metacarpophalangeal joints and hyperflexion of the proximal and distal interphalangeal joints. In children this deformity can be tolerated for a long time without significant joint contracture and



Fig. 10-18 A typical dorsal-burn claw hand. **A-C**, Dorsal burn with hyperextension of the metacarpophalangeal joint and hyperflexion of the proximal and distal interphalangeal joints. **D** and **E**, Results after release with skin graft.

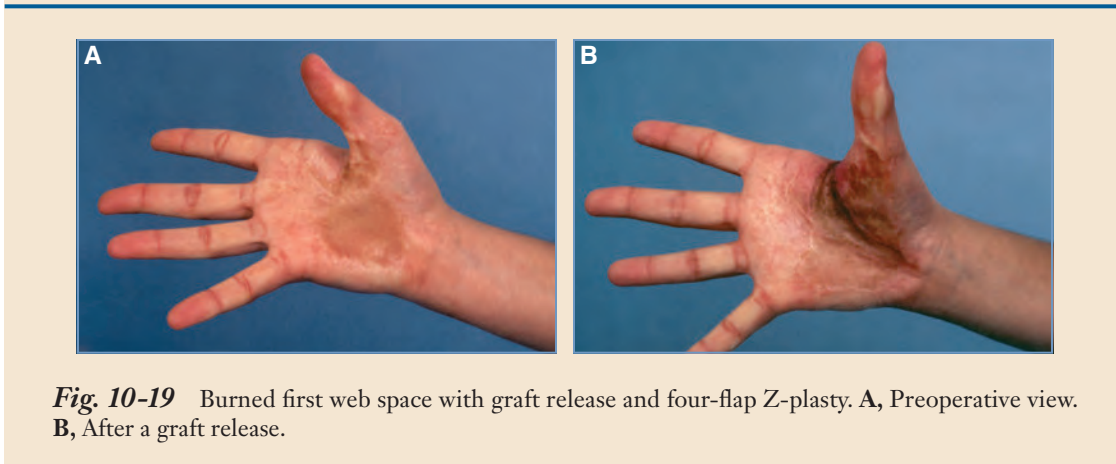


Fig. 10-19 Burned first web space with graft release and four-flap Z-plasty. **A**, Preoperative view. **B**, After a graft release.



Fig. 10-19, cont'd C, Markings for a four-flap Z-plasty. D, Flaps are transposed. E, Long-term result.



Fig. 10-20 Multiple web releases with four-flap Z-plasty to the first web and rotation flaps to other webs. A and B, Markings for a four-flap Z-plasty and rotation flaps. C, Flaps rotated into position. D, Long-term result.



Fig. 10-21 Typical dorsal webbing. **A**, Preoperative view. **B**, Result after release using a full-thickness skin graft.

associated tendon shortening. Correction requires excision of scarred skin to restore transverse and longitudinal hand arches, preservation of the underlying peritenon, internal fixation of the metacarpophalangeal joints at 80 degrees and of the interphalangeal joints at 180 degrees, and coverage with thick sheet grafts. With joint contracture and shortening of the musculotendinous unit or exposure of joints or tendons, flap coverage is necessary for effective treatment.

Although surgeons can perform open releases of the joints and lengthen tendons, the trend is toward excision of the burn scar contracture, coverage with a flap (typically a groin or abdominal flap), and placement of an adjustable external fixator, followed by gradual distraction of the shortened deep structures for necessary lengthening.^{43,44} This can be done for either dorsal or palmar contractures.

Digital contractures are common sequelae of hand burns involving the dorsum of the hand and digits.⁴⁵ Boutonnière deformities occur easily, because the dorsal skin over the proximal interphalangeal joints that protects the central slip is thin. A full-thickness burn in this area immediately exposes the central slip, allowing it to become necrotic and exposing the underlying joint to infection. In severe burn cases these joints are often overlooked because the focus is on lifesaving and resurfacing issues. Ideally, surgeons should perform early debridement followed by flap reconstruction to preserve the underlying tendon and joint structures.^{46,47} However, this is usually not practical, except for isolated burn injuries. Typically, surgeons see chronic burn boutonnière deformities in the convalescent period. Treatment depends on the condition of the proximal interphalangeal joint, and radiographs should be obtained. Fixed deformities and the fingers with joint destruction are best treated with fusion of the proximal interphalangeal joint.⁴⁸ If a finger has normal range of motion and the joint is normal, the surgeon can attempt correction with reattachment of the central slip and centralization of the lateral bands. This usually requires antecedent reconstruction of the joint's dorsal surface with a flap. Interdigital contractures (burn syndactyly) rarely cause significant functional deficits unless they involve the first web space. Z-plasties are associated with lowest recurrence of syndactyly, whereas skin grafts and flaps have a higher rate.³⁸

The thumb represents about 50% of the function of the hand. Burn injuries can result in complete or partial loss of digits and compromised function. Reconstruction of the thumb is the



Fig. 10-22 Toe-to-hand transfer to reconstruct a thumb. **A-C**, Postoperative result. **D**, Donor defect.

basis for successful rehabilitation of the hand. Concerns in thumb reconstruction include preservation of length, mobility, sensibility, and stability. During the acute period, care is needed to preserve the first web space and prevent contracture and palmar adduction. Burn injuries often result in the loss of digits or portions of digits (length), which causes problems with pinch, grasp, and opposition. With loss of thumb and digital length, a number of options are available to restore function. Treatment of web space contractures has already been discussed, but the first web may also have to be deepened to provide mobility and opposition. Phalangization involves deepening the first web by resecting the second metacarpal (with the loss of the index finger).³⁸ This effectively converts the thumb metacarpal and its residual phalanx into a phalanx, allowing some degree of pinch and opposition. If a residual proximal phalanx is present on the index finger, it can be transferred on its neurovascular pedicle to the thumb to lengthen the thumb. This procedure is known as an *on-top-plasty*, which works especially well if the thumb has been lost at the metacarpal phalangeal level.⁴⁹ In rare circumstances with total loss of the thumb with preservation of the index finger, a formal pollicization can be performed.⁵⁰ However, severe injuries are more likely to require a toe-to-hand microvascular transfer⁵⁰ (Fig. 10-22). These can be singular (classic great toe or second toe) or multiple (great toe for the thumb and second toe for a digit to oppose). Preoperative planning may require release of the first web and adequate soft tissue to cover the base of the thumb. Usually a groin flap is placed to accomplish these objectives. Stability issues are typically corrected with joint fusion. Complex reconstructions require adequate healthy soft tissue, which is usually at a premium.

Nail and nail bed injuries can be particularly difficult to treat.⁵¹ Burn injuries destroy and distort the germinal matrix, resulting in painful nail remnants buried in scar tissue. These can be

difficult to eradicate. Treatment is excision of the remaining matrix, but this can be elusive. Another common problem is scar causing retraction of the eponychial fold. This can be painful and cause distortion of the nail bed. Improvement occasionally can be accomplished by releasing the retracted eponychium and placing a skin graft behind the fold. More severe cases may require an advancement or rotation flap of the dorsal or lateral skin to replace the cutaneous portion of the fold and grafting the donor site.⁵²

Lower Extremities

The lower extremities constitute more than 30% of the entire body surface area. The reconstructive approach of the lower extremity is based on the amount and depth of tissue destroyed and follows general principles that apply elsewhere in the body. The pediatric burn population requires special planning for continued growth and probably repeated surgical intervention.

Contractures are most likely to occur across joints (for example, the knees, ankles, and toes). The most common lower extremity deformities that require correction in burn patients are dorsal foot extension contracture, popliteal flexion contracture, and hip flexion contractures.

Scar contracture of the dorsum of the ankle should be released and covered with a split-thickness skin graft. Long-standing contracture can cause shortening of the extensor tendons. Contracture of the posterior ankle universally involves the calcaneal tendons. The use of local flaps in the burned lower extremity is challenging when local donor tissue is limited (Fig. 10-23).

Contracture at the knee is more common than at the ankle. Knee joint contractures, however, are far more easily prevented than those affecting any other lower extremity joint.⁵³ Linear and narrow scar bands may be released and lengthened by a Z-plasty. Wider and thicker scars will have to be released, partially excised, and resurfaced with skin grafts or occasionally by flaps. Reconstruction must be designed so that the desired functional and aesthetic results can be achieved using the simplest method available and with minimal donor tissue or donor site morbidity. To prevent recurrence of contracture, splinting the knee in the extended position is necessary for 2 to 6 months.

For severe injuries that require below-knee amputation, every effort should be made to preserve the knee joint, including the use of free flaps, both soft tissue and osteocutaneous, if length is a problem. In growing children, subsequent growth of the residual tibia may cause repeated ulceration and breakdown, necessitating repeated revisions.



Fig. 10-23 Dorsal ankle and toe release. **A**, Immediately following ankle release after a dorsal toe release. **B**, Final result.

One of the most common problems seen in children is dorsal toe contractures from burns of the dorsum of the foot. Treatment involves release, pinning the toes, and coverage with a skin graft. The pins are removed at 7 to 10 days, and the foot is placed in a shoe with metatarsal bars. Pressure is applied using garments with foam or silicone inserts. Reconstruction of the plantar surface remains a challenge. The unique nature of both plantar and palmar skin, with fibrous connections to deeper structures, prevents shifting of the overlying skin and enhances stability. Loss of this unique property and loss of the associated sensation in weight-bearing areas can cause breakdown, ulceration, and exposure. Although free flap reconstruction may be necessary for deep dermal burns of the plantar surface, it will lack stability and be prone to breakdown and ulceration. Every effort should be made to preserve the weight-bearing portions of the foot.

Genitalia

Perineal burns and injury to external genitalia can occur from a number of sources. With infants, dipping and scald burns frequently involve the perineum and its structures. Males are at a greater risk and generally experience more problems than females because of the inherent differences in genital structure. In the acute phase, problematic contracture is limited by inserting a Foley catheter, splinting the patient in an abducted position, and conservative management of damaged genital tissue.

Perineal contracture is the most common delayed complication. This can be treated conservatively with the appropriate use of Z-plasties and skin grafts to release bands and resurface.⁵⁴ Females commonly have webbing or banding crossing the labia majora; a good outcome can be achieved with standard reconstruction techniques. Although meatal issues are common in males from both the burn injury and the Foley catheter, they rarely occur in females.^{41,42} Loss of the penis, scrotum, testicles, or any combination is possible. The testicles can retract, and combined with subsequent scarring, can appear to be cryptorchid. Scrotums can be reconstructed by bringing down the testicles, pexing them together, and covering them with skin grafts. This is preferable to burying them in the medial thigh, because spermatogenesis takes place below body temperature. Full-thickness burns to the penile shaft can be resurfaced using split-thickness grafts, preferably from a non-hair-bearing area, and appropriate splinting.^{42,55} With total loss of the penile shaft, reconstruction usually requires a free tissue transfer, typically a radial forearm flap (Fig. 10-24).

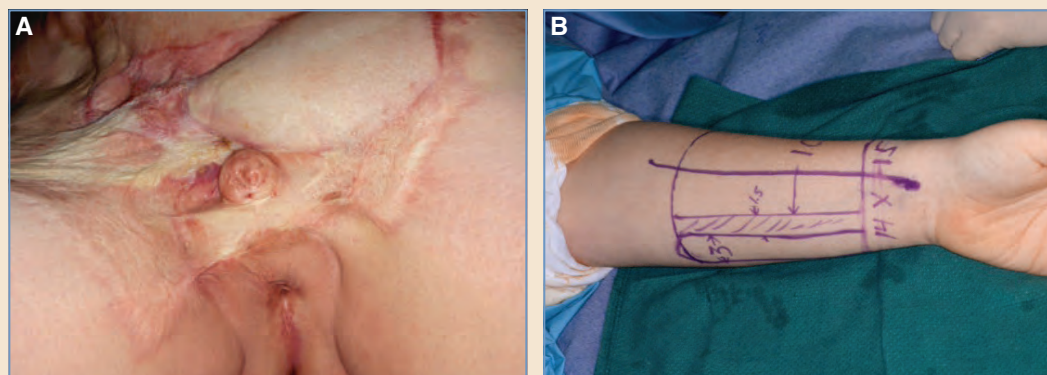
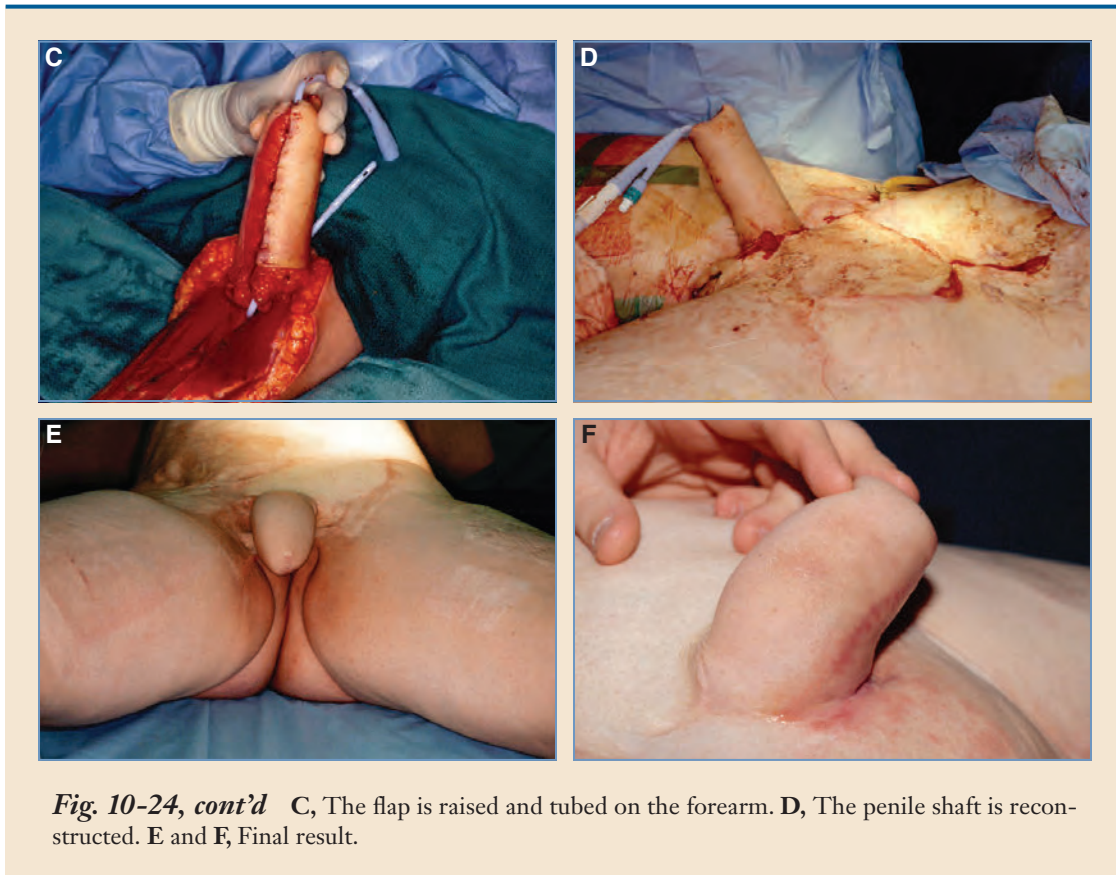


Fig. 10-24 Loss of penile shaft after a muffler burn. **A**, Preoperative defect. **B**, The plan is outlined on the forearm.

Continued



Scar Band Release

Scar bands are sequelae of healing burns that can cause disfigurement and restriction of motion and/or function. In children they tighten during periods of rapid growth. Scar bands that are candidates for release are those that remain tight beyond the growth spurt, especially if they result in a measurable decrease in range of motion. To determine whether a scar release will require subsequent grafting and local or distant flaps, the anatomic site, adjacent tissue, and degree of contracture are considered. The sequence of treatment for scar bands should be managed proximally to distally (for example, the axilla, then the elbow, wrist, hand, and fingers). Contractures can be simultaneously released at multiple levels during the same procedure. Combinations involving skin grafts and local flaps provide versatility in managing scar bands. Scar bands in joint lines are especially well managed with Z-plasty flaps.³

When a scar band release is planned, it is almost always necessary to carry the release incision into the adjacent unscarred skin to achieve adequate improvement in excursion. Postrelease grafts require splinting and pressure treatment, similar to acute burn grafting. A burn therapist should be consulted preoperatively to ensure the postoperative therapy goals reflect the surgical plan. Scar contracture releases that expose joints, tendons, or critical structures will require coverage with distant, pedicled, or free flaps, especially in the hand.

SOCIETAL REENTRY AND SCHOOL INTEGRATION

The period of reentry into society and school is critical for children who have had reconstruction of burn injuries. Not only do they have additional physical and medical needs, but they also have significant psychological requirements. These profound needs and changes can be best addressed with a coordinated reentry program. The burn team should meet with both the parents and school staff to discuss the child's special needs. With appropriate communication, preparation, and education, the process will be greatly eased. The need for continuing medical needs, wound care, splints, and garments should be emphasized. Appropriate precautions and specific methods for dealing with blisters, cuts, and minor wounds should be instituted with teachers and school health staff. The need for ongoing physical therapy and medical appointments may lead to missed classes. It is important to educate classmates and teachers about the child's appearance and injuries. This can preempt derogatory comments or questionable behavior by the students. Visual aids are helpful to explain the injury and its treatment, and time should be set aside to answer questions and concerns. School counselors should be heavily involved to help the child and his or her peers.

KEY POINTS

- Of the 6.6 million major burn injuries that occur each year, approximately half occur in children.
- The most vulnerable pediatric age group susceptible to burn injury is children younger than 5 years of age. Scald, contact, and electrical injuries are most common among children.
- It is estimated that about 20% of pediatric burns are the result of abuse or neglect.
- The potential for hypertrophic or keloid scarring is largely unpredictable; however, there are many risk factors for abnormal scarring in a burn injury.
- The most reliable and proven modality for preventing or treating abnormal burn scars is the use of pressure.
- Improved survival from major burns has led to increased and more complex secondary burn deformities that can significantly affect aesthetics and function.
- The reconstructive approach of the lower extremity is based on the amount and depth of tissue destroyed and follows general principles that apply elsewhere in the body.
- Reentry into society and school is critical for children who have burn injuries.

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11

Vascular Anomalies

Arin K. Greene • John H. Phillips



ascular anomalies are disorders of the endothelium that can affect capillaries, arteries, veins, or lymphatics. They are common, affecting approximately 5.5% of the population.¹ Vascular anomalies are confusing, because numerous types of them exist, lesions often look similar, and imprecise terminology is used.² Vascular anomalies are classified biologically, based on their clinical behavior and cellular characteristics³ (Table 11-1). Using the biologic classification, 90% or more of lesions can be diagnosed by history and physical examination. There are two broad types of vascular anomalies: tumors and malformations.⁴ Tumors demonstrate endothelial proliferation and affect approximately 5% of the population. There are four major types of tumors⁵:

1. Infantile hemangioma
2. Congenital hemangioma
3. Kaposiform hemangioendothelioma (KHE)
4. Pyogenic granuloma

Patients with problematic vascular anomalies are best managed in an interdisciplinary vascular anomalies center.

Table 11-1 Classification of Vascular Anomalies

Tumors	Malformations		
	Slow-Flow	Fast-Flow	Overgrowth Syndromes
Infantile Hemangioma	Capillary Malformation	Arterial Malformation	CLOVES
PHACES association LUMBAR association	CLAPO Cutis marmorata Cutis marmorata telangiectatica congenita Diffuse capillary malformation with overgrowth Fading capillary stain Heterotopic neural nodule Macrocephaly–capillary malformation	Aneurysm Atresia Ectasia Stenosis	KTS Maffucci Parkes Weber Proteus Sturge-Weber
Congenital Hemangioma	Lymphatic Malformation	Arteriovenous Malformation	
Rapidly involuting congenital heman-gioma	Macrocystic and microcystic combined (macrocystic/microcystic) primary lymph-edema Gorham-Stout disease Generalized lymphatic anomaly Kaposiform lymphangiomatosis	Capillary malformation–arteriovenous malformation Hereditary hemorrhagic telangiectasia PTEN-associated vascular anomaly Wyburn-Mason syndrome	
Kaposiform Hemangioendothelioma	Venous Malformation		
	Blue rubber bleb nevus syndrome Cerebral cavernous malformation Cutaneomucosal venous malformation Diffuse phlebectasia of Bockenheimer Fibroadipose vascular anomaly Glomuvenous malformation Phlebectasia sinus pericranii Verrucous venous malformation		
Pyogenic Granuloma			
Rare Vascular Tumors			
Angiosarcoma cutaneovisceral angiomatosis with thrombocytopenia Enzinger intramuscular hemangioma Epithelioid hemangio- endothelioma Infantile myofibroma Tufted angioma			

CLAPO, Capillary malformation of the lower lip, lymphatic malformation of the face and neck, asymmetry of face and limbs, and partial or generalized overgrowth; *CLOVES*, congenital, lipomatous, overgrowth, vascular malformations, epidermal nevi, spinal/skeletal anomalies and/or scoliosis; *KTS*, Klippel-Trenaunay syndrome; *LUMBAR*, lower body hemangioma and other cutaneous defects, urogenital anomalies, ulceration, myelopathy, bony deformities, ano-rectal malformations, arterial anomalies, renal anomalies; *PHACES*, posterior fossa: brain malformations that are present at birth, hemangioma on the skin of the head/neck (>5 cm), arterial lesions: abnormalities of the blood vessels in the head/neck, cardiac abnormalities/aortic coarctation, eye abnormalities, and sternal defects; *PTEN*, phosphatase and tensin homolog.

Vascular Tumors

Vascular tumors generally demonstrate rapid growth, endothelial and angiogenic proliferation, and then postnatal regression. Vascular tumors can be differentiated from vascular malformations by various indicators, such as immunohistologic measures and imaging with MRI and ultrasonography with color flow Doppler imaging.

The International Society for the Study of Vascular Anomalies (ISSVA) classification of vascular tumors divides them into three broad categories: (1) benign, (2) locally aggressive, and (3) malignant.⁶ Benign vascular tumors include infantile hemangiomas, congenital hemangiomas (which are further divided into rapidly involuting [RICH], noninvoluting [NICH] and partially involuting [PICH]), tufted angiomas, spindle cell hemangiomas, epithelioid hemangiomas, and pyogenic granulomas. Locally aggressive vascular tumors include Kaposiform hemangioendothelioma (KHE) and Kaposi sarcoma. Malignant vascular lesions include angiosarcoma (Box 11-1).

Box 11-1 ISSVA Classification of Vascular Tumors

Benign Vascular Tumors

- Infantile hemangioma/hemangioma of infancy
- Congenital hemangioma
 - RICH*
 - NICH
 - PICH
- Tufted angioma
- Spindle-cell hemangioma
- Epithelioid hemangioma
- Pyogenic granuloma (also known as *lobular capillary hemangioma*)
- Others

Locally Aggressive or Borderline Vascular Tumors

- KHE†
- Retiform hemangioendothelioma
- PILA, Dabska tumor
- Composite hemangioendothelioma
- Kaposi sarcoma
- Others

Malignant Vascular Tumors

- Angiosarcoma
- Epithelioid hemangioendothelioma
- Others

*Some lesions may be associated with thrombocytopenia and/or consumptive coagulopathy.

†Many experts think that KHEs are part of a spectrum rather than distinct entities.

NB: Reactive proliferative vascular lesions are listed with benign tumors.

ISSVA, International Society for the Study of Vascular Anomalies; KHE, Kaposiform hemangioendothelioma; NICH, noninvoluting congenital hemangioma; PICH, partially involuting congenital hemangioma; PILA, papillary intralymphatic angioendothelioma; RICH, rapidly involuting congenital hemangioma.

HEMANGIOMAS

Vasculoproliferative tumors are thought to result from the formation of primitive blood vessels from angioblasts. Progenitor cells can be identified in the hemangioma that may originate from the placenta. During proliferation, cellular proliferative factors such as vascular endothelial growth factor (VEGF) are elevated and return to normal levels on involution.⁷⁻⁹ During involution, an increase in the number of mast cells has been seen,¹⁰⁻¹² and mesenchymal stem cells may differentiate into adipocytes in the lesions.^{9,13} During this phase, capillaries undergo apoptosis, which results in resolution.

The expression of glucose transporter 1 (GLUT1) isoform protein is unique to placental endothelial cells and infantile hemangiomas. GLUT1 is not found in congenital hemangiomas such as NICH and RICH.^{14,15} Infantile hemangiomas usually present between 2 weeks and 2 months after birth, and they undergo a proliferative phase, whereas congenital hemangiomas are fully formed at birth.^{14,16}

Infantile Hemangiomas

Hemangiomas are the most common vascular anomaly of childhood.^{17,18} Hemangiomas occur in 20% of low-birth-weight premature infants and are 2 to 4.5 times more frequent in females.¹⁴ Hemangiomas occur in 4% to 5% of white infants but are rare in dark-skinned infants.¹⁴ Placental cells and infantile hemangiomas appear to have a similar life cycle, which suggests that hemangiomas may result from embolized placental cells. The increased risk of hemangiomas in women undergoing procedures that may cause trophoblastic embolization and placental disruption supports the theory of the placental origin of hemangiomas^{19,20} (Fig. 11-1).

Most of these lesions in the skin appear during the first month of life; however, those occurring under the skin may not be noticed until several months after birth and may have a reddish-purple hue when they enlarge (Fig. 11-1, *A*).

During the first 9 to 12 months, these lesions undergo a period of rapid proliferation called the *proliferative phase* (Fig. 11-1, *B*). During the proliferative phase, ulceration and bleeding are common problems for parents. Local wound care by family members is supported by medical staff with regular clinic visits. With ulceration, there is risk for infection and secondary scarring. Bleeding usually responds to light pressure and dressing.

After approximately 12 months, infantile hemangiomas begin to involute and often disappear completely (Fig. 11-1, *C*). Involution is usually complete by 4 years of age.²¹

Infantile hemangiomas are most common in the head and neck region (60%) but can occur anywhere on the body.¹⁴ Hemangiomas on the lumbosacral area have increased risk for spinal dysraphism, tethered cord, and pelvic anomalies. There is also the association of hemangiomas with segmental anomalies. The most common is posterior fossa abnormalities, hemangiomas in the fifth cranial nerve region, arterial intracranial anomalies, cardiac anomalies, eye anomalies, and sternal defects (PHACES) association. There is also LUMBAR association, which stands for *l*ower body hemangiomas, *u*rogenital anomalies, *m*yelopathy, *b*ony deformities, *a*norectal and *a*rtial malformations, and *r*enal anomalies (Box 11-2).

Liver hemangiomas are seen in up to 13% of children with skin lesions, making the liver the most common extracutaneous site.^{14,22} These hepatic hemangiomas may be associated with high-output congestive heart failure as a result of the shunting of blood through the vascular lesion. Other problems rarely associated with infantile hemangiomas may include visual obstruction, nasopharyngeal obstruction, or auditory canal obstruction (Fig. 11-2).



Fig. 11-1 A, Infantile hemangioma in facial skin. B, Infantile hemangioma under the skin in subcutaneous tissue. C, Infantile hemangioma shortly after birth resulting in airway obstruction and the need for a tracheostomy. D, Infantile hemangioma resolving at 3 years of age. E, Infantile hemangioma resolving at 8 years of age.

The most common ocular complication of periocular capillary hemangiomas in infants is visual loss as a result of amblyopia. The strikingly high incidence of amblyopia in some periocular infantile hemangiomas may be related to the onset of the tumor within the first few months of life. Animal research models demonstrate severe amblyopia after only 1 to 4 weeks of visual axis occlusion.²³ Case reviews have reported an incidence of 40% to 60% of amblyopia with periocular capillary hemangiomas.²³⁻²⁵ Vision loss can be caused by strabismus, anisometropia, or visual deprivation. Stigmar²⁴ concluded that the longer the visual axis was occluded, the more severe the amblyopia, reporting that 4 of 51 patients developed moderate to severe amblyopia after only 1 month of occlusion.

In 2008 a group of physicians from Bordeaux Children's Hospital in France described an interesting observation regarding several patients with extensive infantile hemangiomas who received concomitant treatment with propranolol for obstructive hypertrophic cardiomyopathy and high cardiac output.²⁶ Although multiple studies have been reported using propranolol in infantile hemangiomas, there is still no consensus on the optimal dosing regimens and long-term safety profiles.²⁷⁻²⁹

Box 11-2 Regional and Diffuse Associations, Including Vascular Neoplasms**PHACES Association***

- Posterior fossa abnormalities
- Hemangiomas in the fifth cranial nerve region
- Arterial intracranial anomalies
- Cardiac anomalies/coarctation of the aorta
- Eye anomalies
- Sternal defects

LUMBAR Association

- Lower body hemangiomas
- Urogenital anomalies and ulceration
- Myelopathy
- Bony deformities
- Anorectal and arterial malformations
- Renal anomalies

Maffucci Syndrome

- Multiple enchondromas and spindle cell hemangioma
- Hemangioendotheliomas

*Diagnosis requires three or more of these symptoms, one of which must be hemangioma.

Fig. 11-2 Infantile hemangioma of the upper eyelid resulting in visual obstruction.



At The Hospital for Sick Children in Toronto, Ontario, Canada, all children with hemangiomas are assessed by the dermatology service. The exclusion criteria for the use of propranolol was a history of asthma, decreased blood glucose levels, seizures, or suspected PHACES syndrome. The dosage is 0.1 mg/kg/dose tid for 3 days, which is increased to 0.3 mg/kg/dose tid for 3 days, then 0.6 mg/kg/dose tid for 3 days, and finally 0.7 mg/kg/dose for 3 days. Then, if no clinical improvement or worsening has occurred at 2 weeks, the dose should be increased every 2 weeks by 0.5 mg/kg/day, up to a total of 3 mg/kg/day.

There is some controversy with respect to what beta blockade may do to the susceptible developing brain of a neonate. A number of studies in humans indicates that prenatal beta blockade

induces long-term neurologic complications, including impaired school performance, cognitive impairment, and psychiatric disorders^{30,31}; however, no studies to date have examined the long-term neurologic effects of acute or chronic beta blockade in infancy.

Presently the Hospital for Sick Children is using nadolol in patients with infantile hemangiomas, because there is thought to be a decrease in potential side effects compared to propranolol. For weeks 0 to 4, the dosage is 0.5 mg/kg/day divided bid. For weeks 4 to 24, the maintenance phase, the dosage is 2 mg/kg/day. Then there is a deescalation phase during weeks 25 to 52 if there is a complete response or no evidence of proliferation by 10% per week for 4 weeks of weaning. If partial response occurs, a 2 mg/kg/day dosage should be maintained and monitored monthly.

After involution, residual changes may occur, such as residual telangiectasia, skin hypopigmentation, atrophic skin changes, residual skin excess, and fibrofatty tissue. A longer age to involution appears to be related to a greater degree of residual changes. Approximately 10% to 20% of patients require some form of treatment for residual changes.

There is some controversy with respect to the timing of surgery for infant hemangiomas. However, waiting until involution is complete or nearly complete ensures the fewest residual changes present at surgery, including the smallest possible scar with minimal blood loss.

Diagnosis and Imaging

The diagnosis of most infantile hemangiomas is based on history and physical examination, with no imaging studies required. Hemangiomas on grayscale ultrasonography are well defined, solid, and homogeneous, with variable echogenicity. With Doppler ultrasonography, hemangiomas are seen to be hypervascular with arterial and venous wave forms (Fig. 11-3, *A*). MRI shows an intermediate signal on T1-weighted images and a bright signal on T2-weighted images with internal flow voids (Fig. 11-3, *B*).

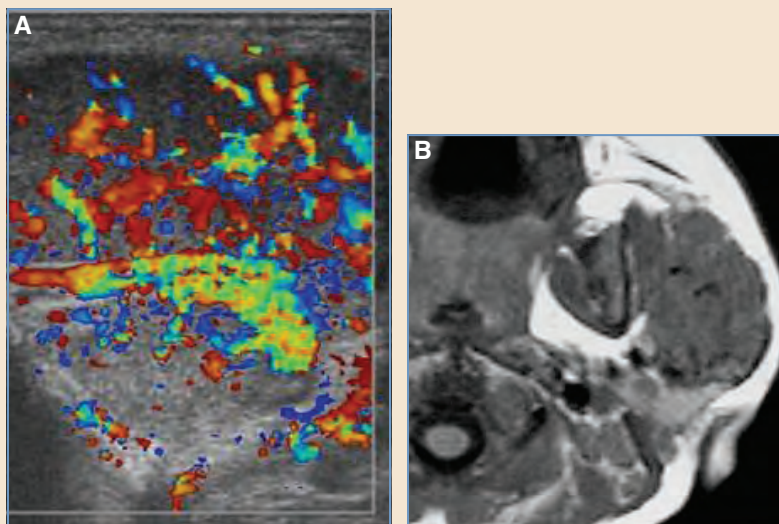


Fig. 11-3 A, Doppler sonogram of an infantile hemangioma in the proliferative phase showing vascular proliferation. B, MRI of infantile hemangioma in the proliferative phase showing vascular filling voids.

Congenital Hemangiomas

Congenital hemangiomas differ from infantile hemangiomas in that they are fully developed at birth and are negative for the GLUT1 protein. Congenital hemangiomas are less common than infantile hemangiomas and there is no sex predisposition.²⁹

There are three main types of congenital hemangiomas:

1. Noninvoluting congenital hemangiomas (NICHs), which are present at birth and then demonstrate proportional growth without regression (Fig. 11-4, *A* and *B*)
2. Rapidly involuting congenital hemangiomas (RICHs), which are present at birth and generally completely regress by 2 years of age²⁹ (Fig. 11-5, *A-D*)
3. Partially involuting congenital hemangiomas (PICHs), which are usually solitary and present in the head and limbs near a joint²⁹ (Fig. 11-6)

On physical examination, congenital hemangiomas may have a pink or violet color with multiple telangiectasias and a surrounding paler halo (Fig. 11-7).

At involution, a RICH may leave residual atrophic skin, but unlike hemangiomas, a RICH rarely leaves residual fibrofatty tissue.³⁰ Some NICHs may partially involute (PICHs), but they do not completely resolve. Imaging findings of congenital and infantile hemangiomas overlap; therefore the radiologist needs to be aware of the clinical history and appearance of the lesion.

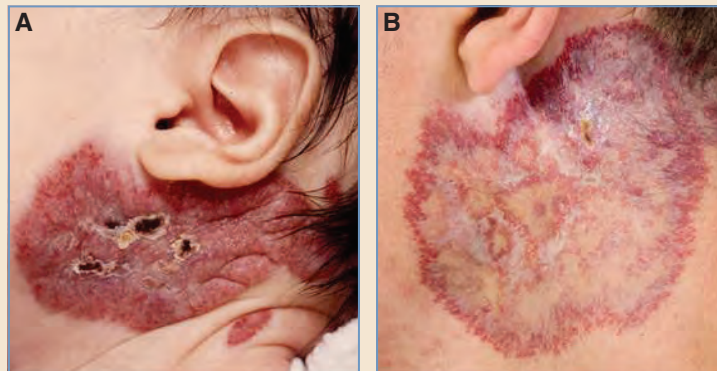


Fig. 11-4 A, NICH as an infant. B, NICH at 18 years of age.

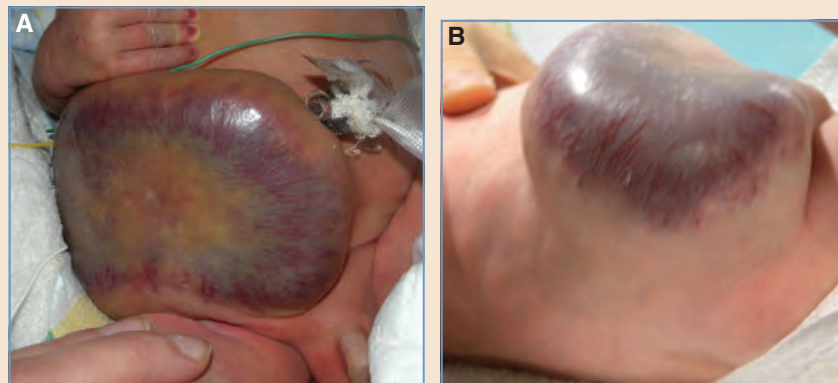


Fig. 11-5 A, RICH at 1 week of age. B, RICH at 6 weeks of age.

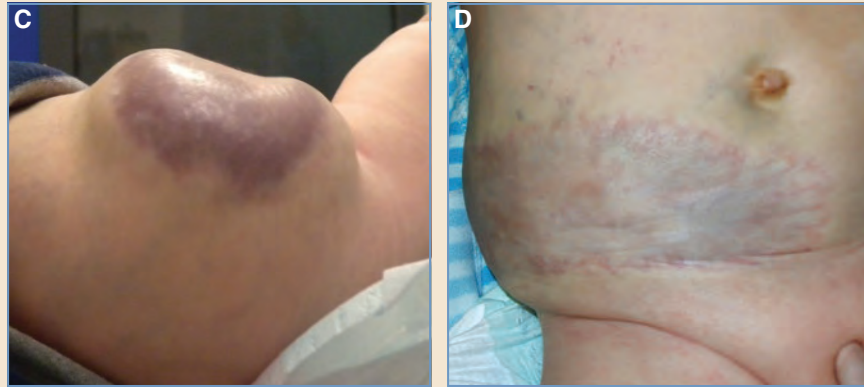


Fig. 11-5, cont'd C, RICH at 3 months of age. D, RICH at 7 months of age.



Fig. 11-6 Congenital hemangioma near joint.



Fig. 11-7 NICH with a surrounding halo.

Fig. 11-8 Pyogenic granuloma on the small finger.



Congenital hemangiomas may have more filling voids, indicative of greater vascularity, and may have calcifications that are not seen in infantile hemangiomas.¹⁷ Treatment of RICHs is similar to that for hemangiomas, although surgery is the treatment of choice for NICHs if clinically warranted.

PYOGENIC GRANULOMAS

Pyogenic granulomas are common acquired benign vasculoproliferative lesions of the skin or mucous membranes. They are most often found in the head and neck region when there is no history of trauma or preexisting skin problems. Pyogenic granulomas are usually solitary and bright red, and they result in bleeding, crusting, and ulceration. They may also be pedunculated (Fig. 11-8).

The cause of pyogenic granulomas is unknown. The following factors have been identified as having a possible role to play in their development:

- Trauma: Some cases develop at the site of a recent minor injury, such as a pinprick.
- Drug-induced: Multiple lesions sometimes develop in patients on systemic retinoids (acitretin or isotretinoin) or protease inhibitors.
- Infection: *Staphylococcus aureus* is frequently present in the lesion.
- Viral infection is possible but not proven.
- The mean age at presentation is 6 to 7 years. Thereafter, there is a decrease in incidence with age.³² Pyogenic granulomas represent 0.5% of skin nodules in children.³³
- Hormonal influences: Pyogenic granulomas occur in up to 5% of pregnancies and are rarely associated with oral contraceptives.
- Pyogenic granulomas are more common in women, because of their frequent formation (5%) on the gingiva during pregnancy.³⁴

Pyogenic granulomas generally require some form of treatment, because they typically do not involute. There are many described treatments for pyogenic granulomas such as CO₂ laser, pulsed dye laser, and silver nitrate application. Surgical excision is the treatment of choice.

TUFTED ANGIOMAS AND KAPOSIIFORM HEMANGIOENDOTHELIOMAS

Tufted angiomas and kaposiform hemangioendotheliomas (KHE) are rare, locally aggressive vascular tumors. Although currently classified as separate entities, they may increasingly be recognized as a spectrum of the same pathology. Tufted angiomas and KHEs are considered neoplasms of intermediate malignancy (ISSVA classification) because of infiltrative growth, local aggressiveness, and variable prognosis.

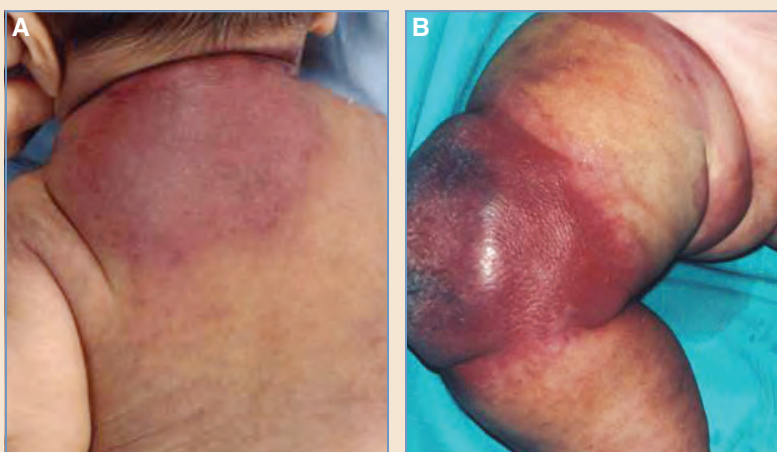


Fig. 11-9 A, KHE on the upper back. B, KHE in the knee area.

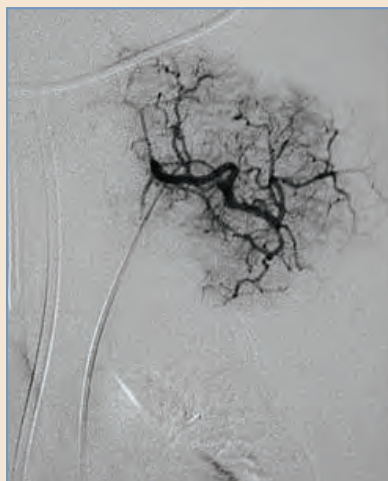


Fig. 11-10 Angiogram of KHE in the upper back demonstrating extensive vascularity.

TAs are rare vasoproliferative tumors that present at or shortly after birth. Histologically, they show vascular tufts of closely packed capillaries in a cannonball pattern.³⁵ These lesions occur on the trunk, extremities, head, neck, and retroperitoneum.³⁶ TAs may be associated with Kasabach-Merritt syndrome with thrombocytopenia as a result of platelet sequestration.³⁷ Surgical excision is the main form of treatment.

KHE also presents at or shortly after birth. These lesions often involve large (greater than 5 cm in diameter) areas on the head, neck, and extremities. They usually appear clinically as flat, reddish-purple edematous lesions (Fig. 11-9).

Histologically, KHE has both vascular and lymphatic components consisting of irregular infiltrating nodules of compressed vessels that may extend into surrounding soft tissues. On imaging, KHE is more often associated with numerous flow voids because of multiple arterial and venous vessels (Fig. 11-10).

Tufted angioma and KHE may also be associated with Kasabach-Merritt syndrome, which if present has a poor prognosis, with up to 30% mortality.³⁸ Surgery is difficult because of the

Fig. 11-11 Kaposi sarcoma of the skin, with red and brown blotches.



involvement of multiple tissue planes and important structures. These lesions may be treated with embolization and then surgical resection where possible; however, surgery is not necessary for lesions that are not causing functional problems. Successful treatment of tufted angioma and KHE has been reported with vincristine.^{31,39,40}

KAPOSI SARCOMA

Kaposi sarcoma is a cancer that develops from the cells that line the lymphatic or blood vessels. Kaposi sarcoma usually appears as tumors on the skin or on mucosal surfaces, such as inside the mouth, but tumors can also develop in other parts of the body such as in the lymph nodes (as bean-sized collections of immune cells throughout the body), lungs, or digestive tract. Kaposi sarcoma is caused by infection with a virus called the *Kaposi sarcoma associated herpesvirus* (KSHV), also known as *human herpes virus 8* (HHV8).^{41,42} The abnormal cells of Kaposi sarcoma form purple, red, or brown blotches or tumors on the skin⁴³ (Fig. 11-11).

The skin lesions of Kaposi sarcoma most often appear on the legs or face. Although they may look painful, they usually cause no symptoms. Kaposi sarcoma can cause serious problems or even become life threatening when the lesions occur in the lungs, liver, or digestive tract. Kaposi sarcoma in the digestive tract, for example, can cause bleeding, whereas tumors in the lungs may cause trouble breathing.

Kaposi sarcoma is rarely seen in children, in whom it is associated with HIV infection and transplant-related immunosuppression.⁴⁴⁻⁴⁸ Endemic Kaposi sarcoma occurs in people living in Equatorial Africa and is sometimes called *African Kaposi sarcoma*. KSHV infection is much more common in Africa than in other parts of the world, so the risk of Kaposi sarcoma is higher. Other factors in Africa that weaken the immune system, such as malaria, other chronic infections, and malnutrition, also probably contribute to the development of Kaposi sarcoma, because the disease affects a broader group of people, including children and women.^{43,49,50}

ANGIOSARCOMA

An *angiosarcoma* is a rare malignant neoplasm characterized by rapidly proliferating, extensively infiltrating anaplastic cells derived from blood vessels and lining irregular blood-filled spaces. Angiosarcomas are aggressive and tend to recur locally, spread widely, and have a high rate of lymph node and systemic metastases. Soft tissue angiosarcoma has a peak incidence in the seventh decade of life, although a wide age range of patients—from 5 to 97 years old—can be affected.⁵¹

Angiosarcomas may occur in any region of the body but are more frequent in skin and soft tissue. Approximately 50% of angiosarcomas occur in the head and neck, but they account for less than 0.1% of head and neck malignancies.⁵² Angiosarcomas also can originate in the liver, breast, spleen, bone, or heart.⁵³⁻⁵⁶

Vascular Malformations

Vascular malformations are errors in vascular development that have minimal endothelial turnover; they affect approximately 0.5% of the population. There are four major categories of vascular malformations based on the anomalous vessel or vessels⁵ (see Table 11-1):

1. Capillary malformation
2. Lymphatic malformation
3. Venous malformation
4. Arteriovenous malformation

Malformations are further divided into rheologically slow-flow and fast-flow lesions.

CAPILLARY MALFORMATION

Clinical Features

Capillary malformation (previously referred to as *port-wine stain*) is the most common type of vascular malformation, affecting 0.3% of newborns.⁵⁷ Capillary malformations can be syndromic and/or associated with underlying structural anomalies. The lesion is noticeable at birth and can involve any area of the integument. The pink-purple skin discoloration can cause psychosocial distress. Over time the lesion progresses, and⁵⁸:

- Darkens
- Fibrovascular cobblestoning can occur
- Pyogenic granulomas may develop
- Soft tissue and bony overgrowth can occur underneath the stain

Phenotypes

Capillary Malformation of the Lower Lip, Lymphatic Malformation of the Face and Neck, Asymmetry of the Face and Limbs, and Partial or Generalized Overgrowth (CLAPO)

CLAPO patients have a midline capillary malformation of the lower lip. The lymphatic malformation is typically microcystic and involves the oral cavity. Patients have normal cognitive development and may have localized or diffuse overgrowth.

Cutis Marmorata

Cutis marmorata is an accentuated pattern of normal cutaneous vascularity common in white infants. Transient mottling occurs with a low temperature that disappears on warming.

Cutis Marmorata Telangiectatica Congenita

At birth the skin of a lower limb in an infant with *cutis marmorata telangiectatica congenita* is depressed, purple, and has a reticulated pattern. Ulceration is common, the leg is often hypoplastic, and iliac/femoral stenosis can occur. Improvement occurs during the first year of life; however, atrophy, pigmentation, and ectasia of the superficial veins often persist.

Diffuse Capillary Malformation With Overgrowth

An extensive extremity capillary malformation can cause soft tissue and bony overgrowth of the limb. The lower extremity is more commonly affected than the upper limb. The extremity has increased circumference and may also have axial overgrowth. Orthopedic consultation is obtained to assess for a leg-length discrepancy.

Fading Capillary Stain

A *fading capillary stain* is a birthmark that is also referred to as an *angel kiss* or *stork bite*. It is present in approximately half of white newborns and is located on the forehead, eyelids, nose, upper lip, or posterior neck. No treatment is necessary, because it lightens over the first 2 years of life.

Heterotopic Neural Nodule

A *heterotopic neural nodule* is a parietal/occipital scalp nodule with overlying alopecia, a surrounding capillary malformation, and a ring of long hair. Lesions contain heterotopic leptomeningeal tissue, and half of these lesions extend intracranially. A neurosurgical consultation is obtained if MRI shows dural involvement.

Macrocephaly–Capillary Malformation

Patients with *macrocephaly–capillary malformation* have the following:

- Macrocephaly
- Developmental delay
- Capillary malformation involving the philtrum/upper lip
- Diffuse capillary malformation involving the trunk or extremities
- Neurologic abnormalities are common

Diagnosis

Diagnosis of capillary malformation is made by history and physical examination; imaging and histopathologic assessment are unnecessary. Lesions affect the skin, are present at birth, and slowly darken.

Management

The mainstay of treatment is pulsed-dye laser (595 nm wavelength). This modality improves the appearance of the lesion by lightening its color⁵ (Fig. 11-12). Intervention during infancy or early childhood is recommended, because:

- Superior lightening of the lesion is achieved.
- The risk of darkening and hypertrophy is reduced.
- Psychosocial morbidity is minimized.

Multiple treatments, spaced 6 weeks apart, are often required until the lesion stops making improvement. Pulsed-dye laser is less effective for capillary malformations that have progressed to a dark color with cutaneous thickening.

Capillary malformations typically do not require surgical intervention. Surgical procedures are indicated to correct overgrowth caused by the malformation⁵ (Fig. 11-13). Patients with facial lesions and soft tissue/bony overgrowth undergo labial reduction, excision of localized cutaneous growths, malar contouring, and/or orthognathic correction. Small fibrovascular nodules or pyogenic granulomas can be excised. Trunk or extremity capillary malformation associated with increased subcutaneous adipose tissue can be improved through suction-assisted lipectomy.

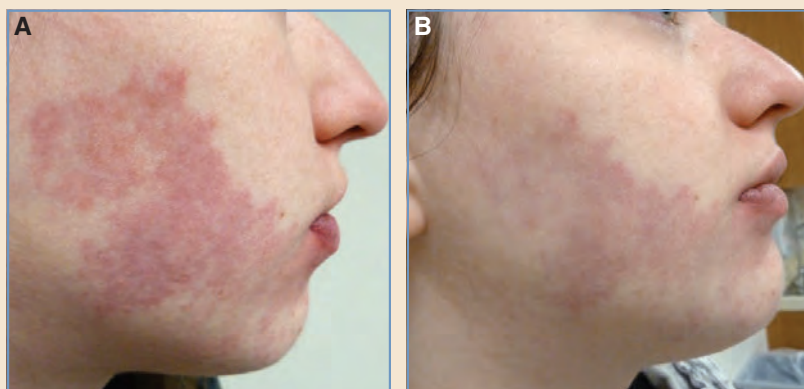


Fig. 11-12 Management of capillary malformation using pulsed-dye laser. **A**, This 20-year-old woman had a capillary malformation involving the right face. **B**, The lightened appearance of the lesion after two treatments. (Courtesy of Sheilagh Maguiness, MD.)



Fig. 11-13 Surgical management of capillary malformation. **A**, This 13-year-old girl had a capillary malformation of the upper lip and cheek causing labial overgrowth. **B**, She had improved lip contour 2½ months after transverse mucosal excision of the hypertrophied tissue.

Severe cutaneous thickening and cobblestoning may be resected and reconstructed through linear closure, skin grafts, or local flaps. If an occlusion cannot be corrected with orthodontics, an orthognathic procedure is considered after completion of skeletal growth.

LYMPHATIC MALFORMATION

Clinical Features

Lymphatic malformation is defined by the size of its channels: macrocystic, microcystic, or combined. Lesions are usually noted at birth, although small or deep lymphatic malformations may not become evident until childhood or adolescence after they have enlarged and/or become symptomatic. The most commonly affected sites are the neck and axilla. Lymphatic malformation causes three major problems: psychosocial morbidity, infection, and bleeding.⁵⁹ Intralesional

bleeding results in pain and swelling. Cutaneous vesicles may have malodorous drainage. Oral lesions can cause macroglossia, pain, poor oral hygiene, and caries. Thoracic or abdominal lymphatic malformations may result in pleural, pericardial, or peritoneal chylous effusions. Periorbital lesions can lead to proptosis, ptosis, amblyopia, reduced vision, and blindness. Intestinal lesions may cause malabsorption. Cervicofacial lymphatic malformations often require tracheostomy to maintain the airway. Osseous lymphatic malformations can cause bone destruction or overgrowth.

Phenotypes

Macrocystic Lymphatic Malformation

Macrocystic lesions contain cysts large enough to be accessed by a needle (typically 5 mm or larger) and are amenable to sclerotherapy. The most commonly affected sites are the neck and axilla.

Microcystic Lymphatic Malformation

Microcystic lesions have cysts that are too small to be cannulated by a needle (usually less than 5 mm) and thus cannot be treated by sclerotherapy. These lesions commonly affect the face and extremities and are often associated with cutaneous vesicles that can bleed and leak lymph fluid.

Combined Lymphatic Malformation

Approximately half of lymphatic malformations are not purely macrocystic or microcystic; rather, they contain both macrocysts and microcysts. The greater the macrocystic composition of the lesion, the better the prognosis, because the lesion can be treated with sclerotherapy.

Primary Lymphedema

Primary lymphedema typically results from hypoplastic lymphatic development in an extremity. The condition usually presents in infancy or adolescence and most commonly involves a lower limb. Several germline mutations can cause primary lymphedema, such as those in *VEGFR3*, *FOXC2*, *SOX18*, and *CCBE1*.

Gorham-Stout Disease

Gorham-Stout disease is a progressive lymphatic anomaly that causes osteolysis of bone. Over time the bone resorbs and causes pain, pathologic fractures, and significant morbidity. Bony changes exhibit progressive osteolysis with resorption and cortical loss. The ribs are most commonly affected, followed by the cranium, clavicle, and cervical spine. Lesions have an associated infiltrative soft tissue abnormality at the site of the bone involvement. One half of all patients with Gorham-Stout disease develop pleural effusions and one fourth have splenic and/or hepatic lesions. Management involves interferon and bisphosphonates.

Generalized Lymphatic Anomaly

Generalized lymphatic anomaly is a multisystem disorder affecting noncontiguous areas. Unlike Gorham-Stout disease, osseous lesions show discrete lytic areas/radiolucencies confined to the medullary cavity; progressive osteolysis and cortical loss do not occur. If symptomatic bony destruction is present, patients are given interferon and bisphosphonates.

Kaposiform Lymphangiomatosis

Kaposiform lymphangiomatosis is a variant of a generalized lymphatic anomaly that causes thrombocytopenia and/or tissue hemorrhage. The lesion involves the mediastinum, bone, spleen, and/or retroperitoneum. Treatment includes drainage of symptomatic effusions, pleurodesis,

sclerotherapy, and medical therapy (such as corticosteroids, vincristine, thalidomide, or sirolimus). The prognosis is poor, and the mortality rate is high.

Diagnosis

Ninety percent of lymphatic malformations are diagnosed by history and physical examination. Small, superficial lymphatic malformations do not require further diagnostic workup. Large or deep lesions are evaluated by MRI to⁵:

- Confirm the diagnosis
- Define the type of malformation (macrocytic, microcystic, or combined)
- Determine the extent of the disease

MRI sequences are obtained with fat suppression and gadolinium. On T1 images, cysts are hypointense. After contrast administration, macrocystic lesions only show enhancement of the wall and septa; venous malformations have more diffuse, heterogeneous enhancement.⁶⁰ Lymphatic malformations are more infiltrative than venous malformations. Because lymphatic malformations have a high water content, they are hyperintense on T2 sequences. Macrocystic lymphatic malformations often have fluid levels because of intracystic blood or proteinaceous fluid. Compared with macrocystic lesions, microcystic lymphatic malformations show more ill-defined borders and slightly greater enhancement. Ultrasonography can provide diagnostic confirmation, document intralesional bleeding, and differentiate between macrocystic and microcystic lesions. Histologic confirmation of lymphatic malformation is rarely necessary.

Management

A lymphatic malformation is benign, and thus intervention is not mandatory.⁵ Small or asymptomatic lesions may be observed. Intralesional bleeding is treated conservatively with pain medication. Patients with more than three infections in 1 year are given prophylactic antibiotics. Recently, oral pharmacotherapy using sirolimus has shown promising results for the treatment of severe microcystic lymphatic malformations. Intervention for lymphatic malformation is reserved for symptomatic lesions or large, asymptomatic macrocysts. Large macrocystic lesions are usually treated prophylactically with sclerotherapy before they bleed or become infected, which can make the lesion microcystic and no longer amenable to sclerotherapy. If possible, intervention should be postponed until after 12 months of age, when the risks associated with anesthesia are lower.⁵ Intervention for a lesion causing a visible deformity should be considered before 4 years of age to limit psychological morbidity.⁵

Sclerotherapy

First-line management for a large or problematic macrocystic or combined lymphatic malformation is sclerotherapy^{5,61} (Fig. 11-14). This technique involves aspiration of the cysts and then injection of an inflammatory substance, which causes scarring of the cyst walls and adhesion to each other. Although sclerotherapy does not remove the lymphatic malformation, it effectively shrinks the lesion and alleviates symptoms. Commonly used sclerosants include doxycycline, sodium tetradecyl sulfate, ethanol, bleomycin, and OK-432. Microcystic lymphatic malformations do not respond to sclerotherapy.

Sclerotherapy produces superior results and has lower morbidity compared with resection. Bleomycin causes minimal swelling and is considered for lymphatic malformations in difficult anatomic areas—such as the oral cavity and airway—or for lesions not responsive to other agents. The use of bleomycin has some benefit for microcystic lesions that are not amenable to resection. Small lesions in adolescents or adults may be treated in the office without image guidance.

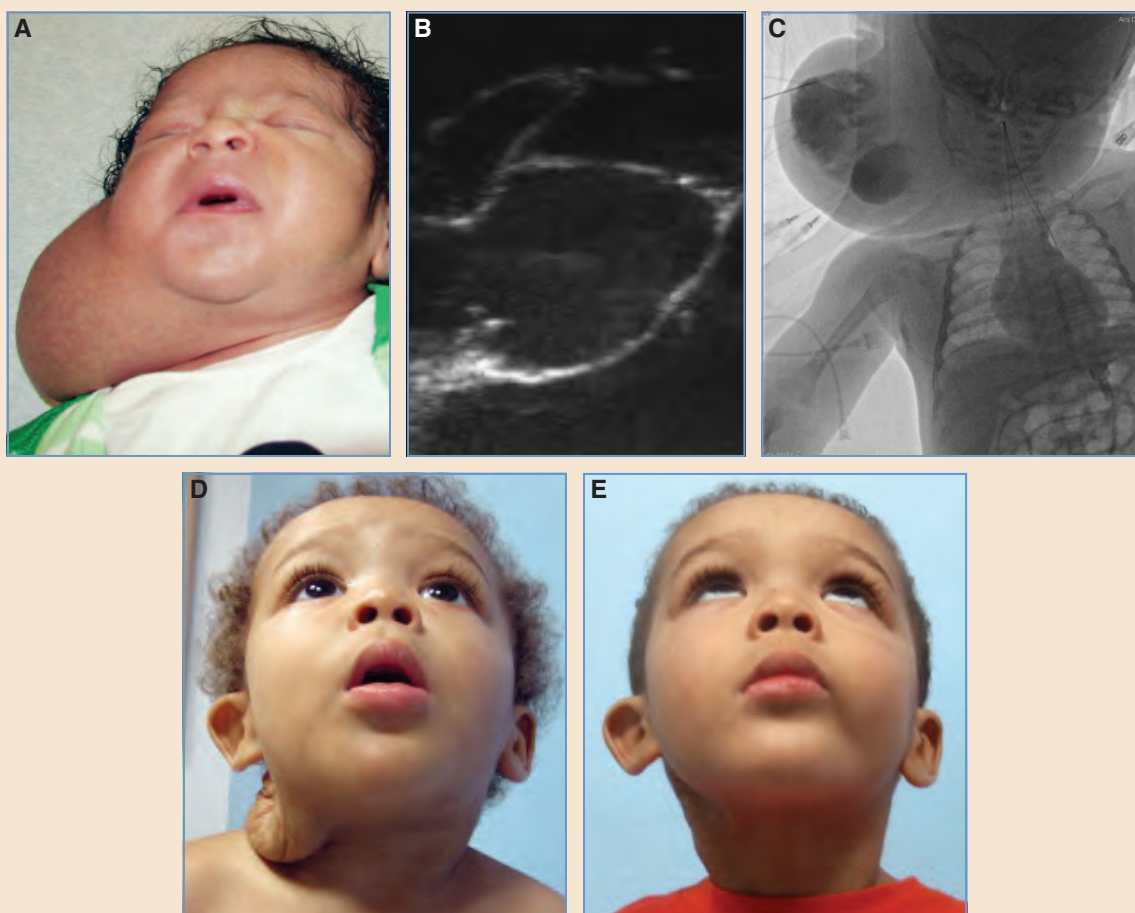


Fig. 11-14 Management of macrocystic lymphatic malformation. **A**, This infant boy had a large lesion involving the neck. **B**, Ultrasound confirmed the presence of macrocysts. **C**, Contrast injection of macrocysts during sclerotherapy. **D**, Two years after sclerotherapy, the child had residual skin excess. **E**, Seven months after resection of redundant skin.

Three percent sodium tetradecyl sulfate is diluted with saline to inject a 1% solution. Most patients, especially children, are managed with a general anesthetic using ultrasonographic and/or fluoroscopic guidance. Large cysts occasionally require placement of a pigtail catheter and sequential drainage and injection over several days.⁵⁹ Additional injections every 6 to 8 weeks may be required. Ulceration is the most common complication of sclerotherapy; it is more likely with superficial lesions and when ethanol is used.

Radiofrequency Ablation

Radiofrequency ablation uses low-temperature tissue destruction that limits thermal injury to the surrounding structures. It is used to treat problematic mucosal vesicles in the oral cavity. Compared with CO₂ laser, radiofrequency ablation provides patients with a faster recovery and less postoperative edema. Reduced swelling is preferable in the oral cavity, because patients can more

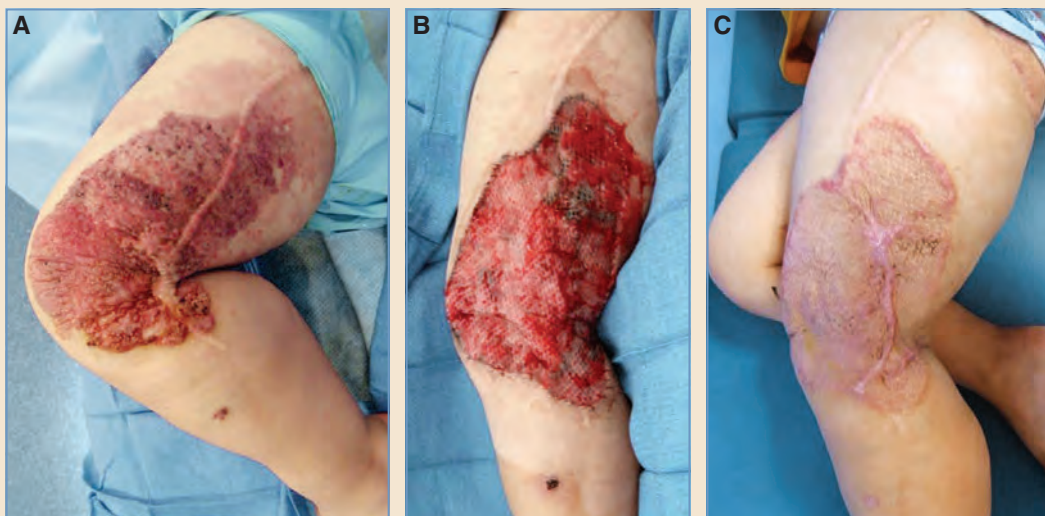


Fig. 11-15 Management of microcystic lymphatic malformation. **A**, This 7-year-old boy had a diffuse microcystic lymphatic malformation of the lower extremity causing bleeding and drainage. **B**, Intraoperative image after resection and split-thickness skin graft placement. **C**, Healed skin graft 6 months postoperatively.

easily resume feeding and there is less risk of airway compromise. The low-frequency mode removes a superficial layer of tissue, and the high-frequency option targets deeper structures.

Resection

Resection of a macrocystic lymphatic malformation generally is not indicated, unless: (1) the lesion is symptomatic and sclerotherapy is no longer possible, because all of the macrocysts have been treated or (2) excision may be curative, because the lesion is small and well localized.⁵ Resection is usually subtotal, because lymphatic malformations typically involve multiple tissue planes and important structures. Nonproblematic microcystic malformations can be observed. Extirpation is reserved for symptomatic lesions, because⁵:

- The excision is usually subtotal
- Recurrence is common
- The scar occurs in the area of the malformation
- The patient can have complications from the procedure

Bleomycin sclerotherapy is often attempted before resection of a microcystic lesion in an unfavorable location. Occasionally, bleomycin can improve the appearance of a facial lesion by 10% to 15% and obviate the need for surgical intervention. When considering resection, the surgeon should ensure that the postoperative scar or deformity after removal of the lymphatic malformation should not be worse than the preoperative appearance of the lesion.⁵ For diffuse lymphatic malformations, subtotal excisions of problematic areas, such as bleeding vesicles or an overgrown lip, should be carried out rather than attempting a “complete” removal, which would result in a worse deformity than the malformation itself.⁵ Most wounds are amenable to linear closure through advancing skin flaps. Skin grafts may be necessary to cover large areas⁵ (Fig. 11-15). Bleeding or leaking vesicles may be treated by CO₂ laser, sclerotherapy, or resection.

VENOUS MALFORMATIONS

Clinical Features

Venous malformations contain veins that are dilated with thin walls and abnormal smooth muscle. Consequently, lesions expand, flow stagnates, and clotting occurs. Although venous malformations are present at birth, they may not become evident until childhood or adolescence when they have grown large enough to cause a visible deformity or symptoms.⁶² Lesions are blue, soft, and compressible. Hard, calcified phleboliths may be palpable. Venous malformations range from small, localized skin lesions to diffuse malformations involving multiple tissue planes and vital structures. Almost all lesions involve the skin, mucosa, or subcutaneous tissue; approximately 50% of lesions also affect deeper structures. Ten percent of patients have multifocal, familial lesions. The primary morbidity of venous malformation is psychosocial, because most lesions affect the skin and cause a deformity. The second most common complication is pain caused by thrombosis and phlebolith formation. Stagnation within a venous malformation causes localized intravascular coagulopathy and thrombosis. Symptoms resolve after the phlebolith is resorbed. Patients with venous malformations are not at risk for thromboembolism, unless a large phlebectatic vein is connected to the deep venous system. Head/neck lesions can result in mucosal bleeding or airway compromise. Venous malformations in the extremities can cause a leg-length discrepancy, hypoplasia, pathologic fracture, hemarthrosis, and degenerative arthritis. Venous malformations involving muscle may result in fibrosis, pain, and disability. Gastrointestinal lesions can cause bleeding and chronic anemia.

Phenotypes

Blue Rubber Bleb Nevus Syndrome

Blue rubber bleb nevus syndrome is a rare, nonhereditary disease with multiple small venous malformations involving the skin, soft tissue, and gastrointestinal tract. Three fourths of individuals with gastrointestinal involvement have bleeding that requires blood transfusions. Resection of intestinal lesions may be required.

Cerebral Cavernous Malformation

Cerebral cavernous malformation is an autosomal dominant disorder with venous malformations involving the brain and spinal cord. The condition results from a mutation in *CCM1/KRIT1*, *CCM2/malcavernin*, and/or *CCM3/PDCD10*.⁶³ Patients are at risk for the development of new intracranial lesions, seizures, and hemorrhage. Nine percent of these patients have a hyperkeratotic vascular malformation of the skin.

Cutaneomucosal Venous Malformation

These small, multifocal mucocutaneous lesions of *cutaneomucosal venous malformation* are caused by a mutation in the *TIE2* receptor; this condition is autosomal dominant. Lesions are small and multiple, and typically affect the skin and oral mucosa, but they also may be located in the muscle, brain, lung, and gastrointestinal tract.

Diffuse Phlebectasia of Bockenheimer

Diffuse phlebectasia of Bockenheimer is an extensive venous malformation affecting all tissues of an extremity. The limb is often hypoplastic. Hemarthropathy causing arthritis and joint destruction can occur. Pain may improve with targeted sclerotherapy to symptomatic areas. Joint disease is managed with arthroscopic resection and/or intraarticular sclerotherapy.

Fibroadipose Vascular Anomaly

Fibroadipose vascular anomaly is a lesion that typically affects the calf, followed by the thigh, forearm, gluteal area, and ankle/foot.⁵ It is differentiated from an intramuscular venous malformation by many characteristics:

- Significant pain
- Contractures
- A nonspongiform venous cutaneous component
- Poor response to sclerotherapy

MRI characteristics of fibroadipose vascular anomaly include:

- Fat/fibrosis
- Minimal brightness on T2 images
- Heterogeneous appearance
- Small, poorly defined channels
- Nonspongiform vessels

Management of fibroadipose vascular anomaly consists of physical therapy, corticosteroid injection, sclerotherapy (only if phlebectasia is present), and/or resection.⁵

Glomuvenous Malformation

Glomuvenous malformation is an autosomal-dominant condition that is caused by a mutation in the *glomulin* gene.⁶³ Lesions are typically multiple, small, and located on the extremities. The lesions are more painful than lesions resulting from a sporadic venous malformation.

Phlebectasia

Phlebectasia involves dilation or varicosity of a vein. There are three forms of an abnormally dilated vein: sporadic, associated with lymphatic malformation, and syndromic, such as a lateral embryonal vein. Problematic phlebectatic veins are usually treated with sclerotherapy, although endovascular laser and excision are other options.

Sinus Pericranii

Sinus pericranii is a venous anomaly of the scalp or face that can have transcalvarial communication with the dura. Before resection of a soft tissue lesion, transcranial veins are obliterated endovascularly to prevent intracranial thrombosis.

Verrucous Venous Malformation

Verrucous venous malformation is a hyperkeratotic venous malformation that has also been referred to as *verrucous hemangioma*. It typically affects the skin/subcutis of the lower extremity. Over time it becomes more hyperkeratotic and bleeds. Treatment is through either CO₂ laser or resection.

Diagnosis

Ninety percent of venous malformations are diagnosed through history and physical examination. Patients and families are queried about a family history of similar lesions. Dependent positioning will cause a venous malformation to enlarge. Small, superficial venous malformations do not require additional diagnostic workup. Large or deep lesions are evaluated by MRI to confirm the diagnosis and define the extent of the malformation.⁶² MRI sequences are obtained with fat suppression and contrast. Lesions are hypointense or isointense on T1 images and hyperintense on T2 sequences.⁶⁰ Phleboliths demonstrate a low-intensity signal. Contrast helps delineate a venous malformation from a lymphatic malformation, because venous lesions enhance uniformly. Morphologically, venous malformation is often circumscribed, lobulated, and isolated to an anatomical structure, usually muscle.⁶² Ultrasonography can be used instead of MRI to image

some localized venous malformations and does not require sedation in young children. Histopathologic diagnosis of venous malformation is rarely necessary.

Management

For patients with a venous malformation on a large extremity compression garments are prescribed to reduce stagnation of blood, phlebolith formation, and pain. Individuals with recurrent discomfort are given low-dose daily aspirin to prevent phlebothrombosis.⁶² Large lesions are at risk for coagulation of stagnant blood and localized intravascular coagulopathy. The chronic consumptive coagulopathy can cause either thrombosis and phlebolith formation or bleeding, such as hemarthrosis.⁶² Venous malformations are not at risk for thromboemboli, because they do not affect the deep venous system, and thrombosed lesions are sequestered from larger veins. However, extensive anomalies of the deep venous system or phlebectasia can cause thromboemboli. Patients at risk for thromboemboli are given low-molecular-weight heparin, and occasionally a vena cava filter is necessary.

Venous malformation is a benign condition in which nonproblematic lesions can be observed. Intervention for a venous malformation is reserved for symptomatic lesions or asymptomatic phlebectatic areas at risk for thromboembolism.⁵ If possible, intervention should be postponed until after 12 months of age, when the risks caused by anesthesia are lowest. Therapy for lesions causing a visible deformity should be considered before 4 years of age to limit psychological morbidity.⁵

Sclerotherapy

Sclerotherapy involves the injection of a sclerosant into the venous malformation, causing cellular destruction, thrombosis, and inflammation. Scarring leads to shrinkage of the lesion. This technique typically is first-line treatment for a problematic venous malformation and is generally safer and more effective than resection.⁶¹ Exceptions to this rule include small and well-localized lesions that can be easily excised for cure, glomuvenous malformations that respond less favorably to sclerotherapy, and venous malformations involving the volar hand or adjacent to an important nerve.⁵ Venous malformations involving the palmar surface of the hand are best treated surgically, because if sclerotherapy were to be performed first, fibrosis could prohibit later surgical intervention, because it would increase the difficulty and risks of the procedure. Sclerotherapy effectively reduces the size of the malformation and alleviates symptoms. Sclerotherapy is continued until symptoms are resolved and/or spaces are no longer available to inject. Often multiple treatments are required, spaced 6 weeks apart. Diffuse venous malformations are managed by targeting specific symptomatic areas. Possible sclerosants include sodium tetradecyl sulfate, ethanol, polidocanol, ethanolamine oleate, or an alcohol solution of zein, bleomycin, and sodium morrhuate. Our center prefers sodium tetradecyl sulfate.⁶² Most patients, especially children, are managed using a local anesthetic with ultrasound and/or fluoroscopic guidance. Small lesions in adolescents and adults may be treated in the office without image guidance. Because sclerotherapy does not remove the malformation, patients can continue to have a deformity after treatment that may be improved by resection.

Resection

Extirpation of a venous malformation can be associated with significant morbidity. Resection should be considered for the following⁵:

- Small, well-localized lesions that can be completely removed
- Persistent symptoms after the completion of sclerotherapy
- Lesions involving the volar hand or near an important nerve in which scarring from sclerotherapy would increase the risks of later surgical intervention

When considering resection, the surgeon should weigh the postoperative scar/deformity after removal of the venous malformation against the preoperative appearance of the lesion. Almost all venous malformations should have sclerotherapy before surgical intervention to facilitate the resection, improve the outcome, and lower the recurrence rate. After sclerotherapy, the venous malformation is replaced by scar tissue; thus the risk of blood loss, iatrogenic injury, and recurrence is reduced.

In addition, fibrosis facilitates resection and reconstruction⁵ (Fig. 11-16). Small, well-localized venous malformations may be removed without preoperative sclerotherapy. Gastrointestinal venous malformations with chronic bleeding are typically managed by resection, although solitary lesions can be treated by endoscopic banding or sclerotherapy. A localized venous malformation may be excised and the wound edges reapproximated without complex reconstruction.

For diffuse malformations, staged resection of defined regions is recommended. Subtotal resections of problematic areas, such as bleeding lesions or an overgrown lip, should be carried out rather than attempting a “complete” excision of a benign lesion, which would result in a worse deformity than the malformation itself.⁵

Patients and families are counseled that venous malformations can expand after the excision; thus additional surgical intervention may be required.

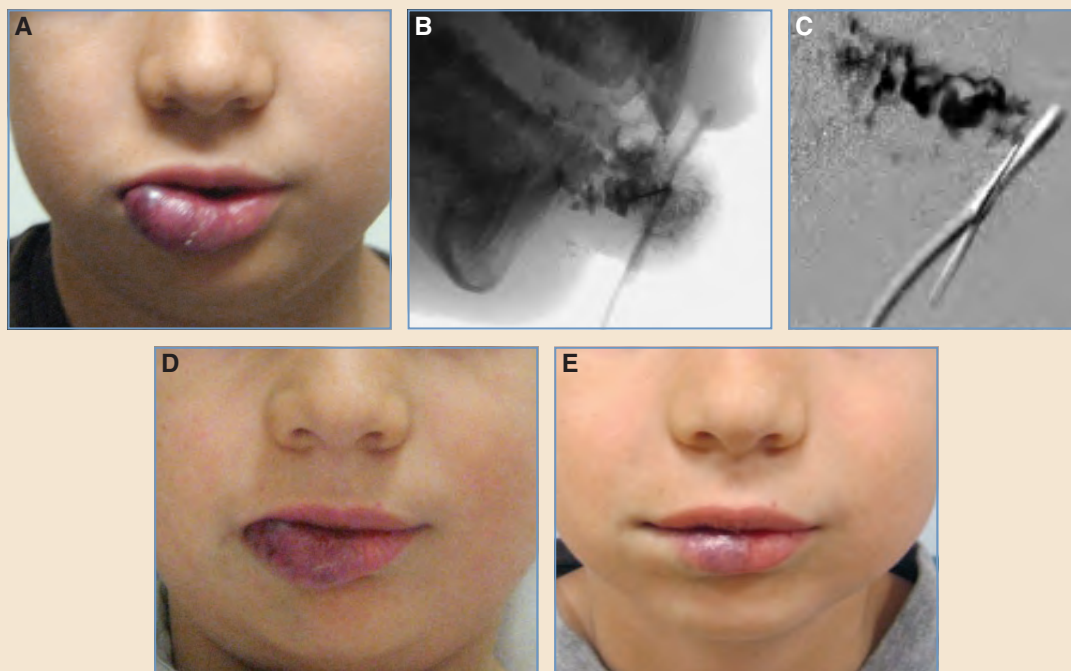


Fig. 11-16 Management of venous malformation. **A**, This 5-year-old boy had a venous malformation of the lower lip. **B** and **C**, Contrast injection of the lesion during sclerotherapy. **D**, He has improved lip contour after three sclerotherapy treatments. Additional sclerotherapy was not possible, because accessible vascular channels had been obliterated. **E**, Six weeks after excision of residual fibrotic venous malformation using a transverse mucosal incision.

ARTERIOVENOUS MALFORMATION

Clinical Features

An *arteriovenous malformation* (AVM) has an absent capillary bed that causes shunting of blood directly from arterial circulation to venous circulation through a *fistula* (direct connection of an artery to a vein) or *nidus* (abnormal channel bridging the feeding artery to the draining veins).⁶⁴ Although present at birth, an AVM may not become evident until childhood, after it has enlarged or become symptomatic.⁶⁵ Lesions have a pink-red cutaneous stain, are warm, and can have palpable pulsations. Arteriovenous shunting reduces capillary oxygen delivery causing ischemia. Patients are at risk for pain, ulceration, bleeding, and congestive heart failure. AVMs also can cause disfigurement, destruction of tissues, and obstruction of vital structures. AVMs progress over time and are classified according to the Schobinger staging system⁵ (Table 11-2).

Phenotypes

Capillary Malformation–Arteriovenous Malformation

Capillary malformation–arteriovenous malformation is an autosomal dominant condition exhibiting atypical capillary malformations that are small, multifocal, round, pinkish-red, and usually surrounded by a pale halo.⁶⁴ Handheld Doppler shows fast-flow. The condition is caused by a mutation in *RASA1*. One third of children with capillary malformation–arteriovenous malformation also have an AVM. MRI of the brain and spine is performed to rule out AVMs in these areas.

Hereditary Hemorrhagic Telangiectasia

Hereditary hemorrhagic telangiectasia is an autosomal dominant condition that is also referred to as *Osler-Weber-Rendu syndrome* and is caused by mutations in endoglin (*ENG*) and activin A receptor type II-like 1 (*ACVRL1/ALK1*). Clinical findings consist of epistaxis, mucocutaneous telangiectasias, visceral AVMs, and a first-degree relative with the condition. Morbidity can include:

- Upper gastrointestinal bleeding
- Stroke and brain abscesses from pulmonary arteriovenous shunting
- Hemorrhage from a cerebral AVM
- High-output heart failure or portal hypertension from a hepatic AVM
- Chronic anemia typically from epistaxis

PTEN-Associated Vascular Anomaly

Patients with *PTEN* mutations have *PTEN hamartoma-tumor syndrome* (previously called *Cowden syndrome* or *Bannayan-Riley-Ruvalcaba syndrome*).⁶⁴ Half of these individuals have a fast-flow vascular anomaly with arteriovenous shunting, referred to as a *PTEN-associated vascular anomaly*.

Table 11-2 Schobinger Staging of Arteriovenous Malformation

Stage	Clinical Findings
I (Quiescence)	Skin warm, pink-blue, shunting on Doppler
II (Expansion)	Enlargement, pulsation, thrill, bruit, tortuous veins
III (Destruction)	Dystrophic skin changes, ulceration, bleeding, pain
IV (Decompensation)	Cardiac failure

(PTEN-AVA).⁶⁴ PTEN-AVAs can be multifocal, contain ectopic adipose tissue, and have segmental dilation of draining veins. Patients with this syndrome exhibit macrocephaly and males have penile freckling. Other features can include developmental delay/autism, thyroid lesions, and/or gastrointestinal polyps. Patients with a PTEN mutation are followed for the presence of tumors, particularly endocrine and gastrointestinal malignancies. PTEN-AVAs are managed similarly to nonsyndromic AVMs.

Wyburn-Mason Syndrome

Wyburn-Mason syndrome is a condition that has also been called *Bonnet-Dechaume-Blanc syndrome* and *retinocephalofacial vascular malformation syndrome*.⁵ The syndrome consists of retinal AVMs with or without brain AVMs and a facial capillary malformation or AVM.

Diagnosis

Ninety percent of AVMs are diagnosed by history and physical examination. Hand-held Doppler examination shows fast-flow. If the diagnosis is equivocal after history, physical examination, and handheld Doppler, ultrasound is the first-line study to confirm the diagnosis. Color Doppler shows a poorly defined hypervascular lesion with fast-flow and shunting; a parenchymal mass is not present. Tortuous feeding arteries are visualized with dilated draining veins and significant flow. MRI is usually obtained to confirm the diagnosis and determine the extent of the lesion. AVMs exhibit dilated feeding arteries/draining veins, enhancement, and flow voids.⁶⁰ Magnetic resonance angiography (MRA) shows feeding arterial vessels and early enhancement of draining veins. An angiogram is obtained if the diagnosis remains unclear after ultrasound and MRI, or if embolization is planned. Angiography shows the flow dynamics of the lesion and identifies the dilated arteries, the enlarged draining veins, and the nidus. The nidus contains tortuous, small vessels with ill-defined vascular spaces. Histopathologic diagnosis of AVM is rarely necessary.

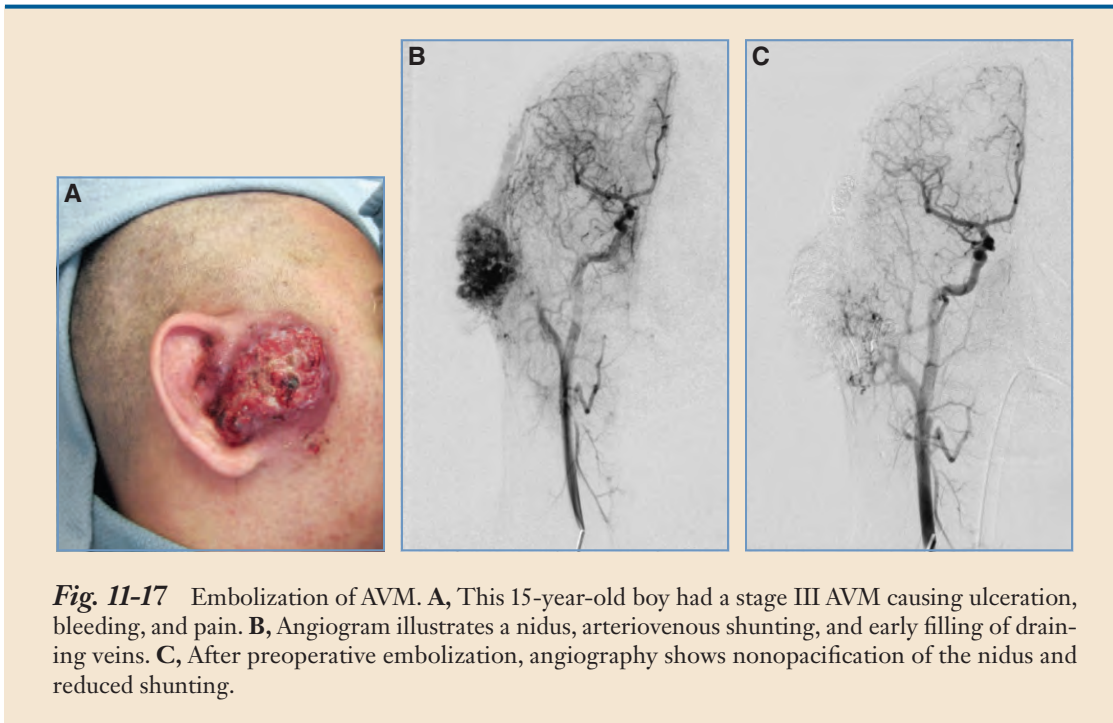
Management

AVM is not a malignancy and intervention is not mandatory.⁵ Management is focused on alleviating symptoms, preserving vital functions, and improving a deformity. Because the lesion is often diffuse and involves multiple tissue planes, a cure is rare. An asymptomatic AVM should be observed unless it can be removed for possible cure with minimal morbidity.⁵ Embolization or incomplete excision of an asymptomatic lesion may stimulate it to enlarge and become problematic. Intervention must be individualized based on the degree of deformity that would be caused by excision and reconstruction.

For example, a stage I AVM in a location that is not anatomically important may be easily resected before it progresses to a higher stage when excision is more difficult and the recurrence rate is greater. In contrast, a large asymptomatic AVM on the face is easily noticeable, especially in a young child who is not psychologically prepared for a major procedure.⁶⁵ Resection and reconstruction may result in a more noticeable deformity or functional problem than the malformation itself. Intervention for a stage II AVM is similar to that of a stage I lesion, although the threshold for treatment is lower if an enlarging lesion is causing a worsening deformity or if functional problems are expected.⁶⁵ Stage III and IV AVMs require intervention to control pain, bleeding, ulceration, or heart failure.

Embolization

Embolization is the delivery of substance through an arterial catheter to occlude blood flow and/or fill a vascular space. Success requires that the embolic agent reaches the nidus of the AVM at



the point of initial venous drainage.⁶⁶ In addition to the replacement of arterialized blood with an inert embolic substance, ischemia and scarring further reduce arteriovenous shunting, shrink the lesion, and improve symptoms. Embolization is not curative, and most AVMs will reexpand after treatment.⁶⁵ Indications for embolization include preoperative intervention to reduce blood loss during resection (Fig. 11-17) or definitive treatment to alleviate symptoms for lesions not amenable to resection.⁵ Proximal arterial embolization is contraindicated, because recanalization occurs and the lesion becomes inaccessible for future embolization. Because the AVM is not removed, almost all lesions eventually will expand after treatment.⁶⁵ Embolization can effectively palliate an AVM by alleviating pain and bleeding. Substances used for embolization may be liquid (N-butyl cyanoacrylate, Onyx, ethanol) or solid (polyvinyl alcohol particles, coils).⁶⁶ The most frequent complication of embolization is ulceration, which is more common for superficial lesions. Distal migration of embolic material can cause ischemic injury to uninvolved tissues.

Sclerotherapy

Sclerotherapy is the transcutaneous injection of a substance into the malformation, which causes endothelial destruction and thrombosis. Indications for sclerotherapy include a well-localized AVM, occlusion of veins draining the AVM, and an AVM that cannot be accessed transarterially, usually because previous embolization has occluded the feeding arteries.⁵ Sclerotherapy of an AVM is riskier than treatment of a slow-flow lesion, because the sclerosant is more likely to escape into the systemic circulation.

Resection

Resection of an AVM has a lower recurrence rate than embolization.⁶⁵ Indications include a well-localized lesion, correction of a focal deformity, or a symptomatic AVM that has failed embolization⁵ (Fig. 11-18). Extirpation and reconstruction of a large, diffuse AVM should be exercised with caution, because cure is rare and the recurrence rate is high, the resulting deformity is often



Fig. 11-18 Resection of AVM. **A**, This 39 year-old-woman had an ulcerated, bleeding, painful stage III AVM of the forehead and scalp since adolescence. **B**, Intraoperative view after preoperative embolization. **C**, Soft tissue defect after resection. **D** and **E**, The wound was closed using a split-thickness skin graft. **F**, Nine months postoperatively, the graft is concealed by hair.

worse than the appearance of the malformation, and resection can be associated with significant morbidity.⁵ When excision is planned, preoperative embolization will facilitate the procedure by minimizing blood loss. Excision should be carried out 24 to 72 hours after embolization, before recanalization restores blood flow to the lesion. Small, well-localized AVMs or those that cannot be accessed for embolization may be resected without preoperative embolization. Proximal feeding vessels to the AVM should never be ligated, because collateralization will stimulate enlargement, and access for future embolization will no longer be possible. Most defects can be reconstructed by advancing local skin flaps. Grafting ulcerated areas from the skin has a high failure rate, because the underlying tissue is ischemic. Excision with a regional flap transfer may be required. Free-flap reconstruction permits wide resection and closure of complicated defects.

VASCULAR MALFORMATION OVERGROWTH SYNDROMES

CLOVES Syndrome

Congenital lipomatosis, overgrowth, vascular malformations, epidermal nevi, and scoliosis/skeletal/spinal anomalies (CLOVES) is a condition that is caused by a somatic mutation in *PIK3CA*.⁵ Many of these patients previously were thought to have Proteus syndrome. Patients with CLOVES do not have cerebriform connective tissue nevi involving the hands and feet, and the condition is not significantly progressive. All individuals with CLOVES syndrome have a truncal lipomatous

mass, a slow-flow vascular malformation, and hand/foot anomalies. Other common features include paraspinal fast-flow malformations, linear epidermal nevus, and hemihypertrophy. MRI is obtained to determine whether the spinal cord is threatened by a lipomatous lesion or AVM. Children are at risk for developing Wilms' tumor and require screening.

Klippel-Trenaunay Syndrome

Klippel-Trenaunay syndrome (KTS) is a capillary-lymphatic-venous malformation of an extremity causing overgrowth.⁵ The condition almost always affects the lower limb. The contralateral foot or hand can be enlarged and exhibit macrodactyly. The venous component of KTS manifests as phlebectasia and abnormal drainage of the affected area. A large embryonic vein in the subcutaneous tissue known as the *marginal vein of Servelle* is isolated in the lateral calf/thigh and communicates with the deep venous system. Complications include thrombophlebitis and pulmonary embolism. The lymphatic abnormalities are usually macrocystic in the pelvis/thigh and microcystic in the abdominal wall, buttock, and distal limb. MRI is obtained to confirm the diagnosis and determine the extent of the disease. An important feature of KTS is that the tissues below the muscle fascia are affected by the malformation. Patients with KTS are not at increased risk for Wilms' tumor; thus screening is unnecessary. Because embryonal veins can connect to the deep venous system causing thromboembolism, these veins are removed in early childhood with sclerotherapy or endovascular laser.

Maffucci Syndrome

Maffucci syndrome is a nonhereditary disorder that is characterized by multiple enchondromas and soft tissue vascular lesions.⁵ The condition is caused by a somatic mutation in isocitrate dehydrogenase. The soft tissue vascular anomaly is called a *spindle cell hemangioma*, but this lesion is likely a reactive process in an underlying venous malformation. The spindle cell hemangioma/venous malformation can affect the hand, foot, arm, leg, trunk, and head/neck. The vascular lesions may cause overgrowth, pain from phlebothrombosis, and limitation of function. Enchondromas most commonly involve the hands. Problematic enchondromas are treated by curettage/excision and bone grafting. Chondrosarcomas are managed based on grade. Soft tissue spindle cell hemangioma/venous malformation can be treated with sclerotherapy and/or resection if symptomatic.

Parkes Weber Syndrome

Parkes Weber syndrome consists of a diffuse AVM of an extremity (usually the leg) causing soft tissue and/or bony overgrowth.⁵ A capillary malformation involves the skin of the affected limb. Parkes Weber syndrome can be sporadic or familial, resulting from a mutation in *RASA1*. Patients have subcutaneous and intramuscular microshunting and can develop congestive heart failure. Most children are observed until symptoms necessitate intervention. Embolization may reduce congestive heart failure, pain, or ulceration. Occasionally, amputation is necessary.

Proteus Syndrome

Proteus syndrome is a very rare sporadic overgrowth disorder caused by a somatic mutation in *AKT1*.⁵ The major features are:

- Progressive overgrowth of body parts (typically skeletal/limbs)
- Cerebriform connective tissue nevi (volar hands, plantar feet, chest)

- Epidermal nevi
- Vascular malformations
- Adipose overgrowth
- Cerebral anomalies
- Ophthalmologic findings
- Cystic lung disease
- Renal/urologic anomalies
- Bone disorders (skull hyperostoses, megaspondylodysplasia)

The significant progression of the disease and the cerebriform connective tissue nevus are pathognomonic for Proteus syndrome. There is a 20% mortality rate among affected individuals from venous thrombosis/pulmonary embolism, cystic lung disease, and neoplasms.

Sturge-Weber Syndrome

Sturge-Weber syndrome is defined by a capillary malformation in the V₁ trigeminal nerve distribution with ocular abnormalities (glaucoma, choroidal vascular anomalies) and/or a leptomeningeal vascular malformation.⁵ Patients also commonly have soft tissue and/or bony overgrowth.⁵⁸ Extensive pial vascular lesions may cause refractory seizures, hemiplegia, and/or delayed cognitive development. Patients also often have extracraniofacial capillary malformations. Any child with a capillary malformation in an upper trigeminal nerve distribution should be screened for Sturge-Weber syndrome. MRI is obtained to rule out leptomeningeal vascular lesions. Patients undergo an ophthalmologic evaluation to assess for choroidal anomalies and glaucoma.

KEY POINTS

- Vascular anomalies are common and affect both children and adults.
- The field of vascular anomalies is confusing, because numerous types of vascular anomalies exist, different lesions have a similar appearance, and the terminology can be confusing.
- Accurate diagnosis of the lesion must be ascertained before treating the lesion.
- Patients with problematic vascular anomalies are best managed in an interdisciplinary vascular anomalies center.
- Vascular malformations are managed by observation, laser, sclerotherapy, embolization, or resection; pharmacotherapy is not available.

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12

Pediatric Tissue Expansion

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issue deficiency is the underlying problem encountered in the care of many congenital and acquired deformities in the pediatric population. With deficiencies in both the soft tissue and skeleton, no single treatment modality can provide all the answers. Experience with a broad spectrum of difficult pediatric reconstructive problems has demonstrated that *tissue expansion* (TE), either as a primary modality or as an adjunct to other reconstructive techniques, provides a reliable mechanism for replacing the missing soft tissues. When treating large skin lesions or scars, TE provides like-quality and like-texture skin to replace the tissue to be excised. When treating craniofacial deformities (either congenital or acquired), TE can provide added tissue to cover the reconstructed skeleton and replace skin deficiencies. TE also provides a means of moving composite tissues from distant sites to provide skin coverage and soft tissue fill after resection of tumors and to reconstruct contour defects where both the size of the donor area and closure of the donor defect present possible limitations.

BACKGROUND

Although the genesis of modern day TE is credited to innovators such as Radovan¹ and Austad and Rose,² the technique takes some of its roots from early lessons in distraction osteogenesis. Bone traction with either internal or external devices at the turn of the twentieth century led to the concept that mechanical stress could lengthen tissue.^{3,4} Putti⁵ extrapolated these ideas from bone to surrounding soft tissue by placing constant tension on a composite tissue to obtain soft tissue lengthening. In the 1950s, Neumann⁶ became the first surgeon to use an expansile implant, when he used a latex balloon to enlarge periauricular skin for a traumatic ear deformity. Despite these early efforts, it was not until 20 years after Neumann's report that TE was revisited. Charles Radovan,¹ a resident at Georgetown, reintroduced the concept of expansion when he inserted a contemporary device with an internally placed port. Shortly thereafter, Eric Austad² produced a

self-inflating device. In 1982 the first National Tissue Expansion Symposium was sponsored by the Plastic Surgery Educational Foundation, marking the recognition of a new advance in reconstructive surgery. Since that time expansion has been applied to many reconstructive problems, with applications in both regional expansion and expansion at distant sites for subsequent graft and flap transfer. A better understanding of expansion has led to many flap design modifications, thereby increasing its worth as a reconstructive option.⁷

PHYSIOLOGY

Skin is a unique substance composed of a viscoelastic cellular and extracellular matrix. When a constant mechanical stress is applied to skin over time, two phenomena have been observed: *mechanical creep* and *biologic creep*. Mechanical creep is based on morphologic changes that occur on a cellular level in response to the applied stress—the cell is stretched. Disruption of gap junctions and increased tissue surface area also result in cell proliferation (biologic creep). Growth of the tissue by cell proliferation restores resting tension of the tissue to baseline.⁸ Histologically, the early work of Sasaki⁹ and his colleagues pointed to a proliferative response of the epidermis with concurrent thinning of the dermis and alignment of collagen fibrils. In humans, these effects are maximized at 6 to 12 weeks after expansion.¹⁰ On a molecular level, a panoply of growth factors, cytokines, hormones, adhesion molecules, cytoskeletal elements, and signal transduction proteins is induced in response to expansion, confirming that TE is a dynamic process.¹¹

One of the features that makes an expanded flap so reliable is its improved vascularity. Many studies have demonstrated that the vascularity of an expanded flap is superior to its nonexpanded counterpart in both the number and caliber of vessels.¹² Moreover, angiogenic factors, such as vascular endothelial growth factor, are expressed on the surface of expanded tissue at a significantly higher level than on nonexpanded controls.¹³ This augmentation of blood flow is attributable to the capsule that forms around the expandable prosthesis. Because expanded flaps and delayed flaps are similar in vessel caliber, TE has been regarded as a form of the delay phenomenon. An expanded flap, therefore, is a delayed flap.

KEY CONSIDERATIONS

Successful expansion, regardless of the indication, begins with careful case selection. The surgeon must have an understanding of the potential problems and how to avoid them, and between the surgeon and his or her nursing staff, the parents and patients (when old enough) must be brought to a thorough understanding of the expansion process and its potential complications.

The choice of expander, the incision for placement, and the expansion routine may vary, depending on the site of expansion and the underlying defect being treated. Experience has demonstrated that a higher rate of complications in nonnevus cases may require variation in technique. Also, a surgeon should always consider whether techniques other than expansion are more suitable for reconstruction in a particular case.

Tissue Expander Design

Several types of tissue expanders exist, based on shape, size, and type of filling valve. Expanders can be standard, customized, anatomic to the donor site (breast), or differential in fill volume to provide tissue tapering. They follow three basic shape patterns: round, rectangular, and crescent (and croissant, a variant of crescent). The more commonly used are the round and rectangular types. Crescent (and croissant) prostheses, developed to minimize dog-ears at the donor site, have



Fig. 12-1 Rectangular tissue expander with a thicker base plate, tubing, and remote filling port.

fallen out of favor, because the added tissue gained with rectangular expanders may increase the potential choices for flap design (such as transposition flaps). Outside of expansion for breast reconstruction, we now use rectangular expanders in all cases.

Expander volumes have a wide range and vary according to the anatomic site. Round expanders of 100 to 2000 cc and rectangular expanders of 70 to 1200 cc are available (smaller and larger custom expanders are available to meet uncommon anatomic requirements). Saline solution is delivered in a controlled fashion through the valve port, which is either integrated into the prosthesis or connected to the device by a customized length of silicone tubing (Fig. 12-1). An integrated system offers the advantage of undermining a single pocket for expander placement, but it also places the implant at risk of perforation by a misplaced needle. On the other hand, internal remote ports remove the danger of perforating the prosthesis itself but introduce the potential complications of flipping over, tube obstruction, and migration. To avoid these complications, the port pocket undermining should be minimal, the port should be placed over firm supporting tissue, and, if necessary, the port position should be fixed with sutures (see Fig. 12-4, *E*). In more mobile areas, such as an extremity, the risk of the port flipping over can be minimized by using a larger (standard) port rather than a “mini” port. The port size does not affect the amount of fill that can or should be provided to various sizes of tissue expanders.

A variety of expanders are available with designs aimed at keeping the profile of the expander low. In our experience, several of these designs gain low profile at the expense of increased firmness of the expander envelope. We currently use an expander with slightly firm backing that allows the redundant envelope to be folded beneath the base when first placed. This avoids folds and firm points that may compromise overlying skin. As the expander increases in size, the redundant envelope gradually unfolds beneath the enlarging flap.

Expander Placement and Expansion Routine

Consideration for incisions, expander placement, flap movement in relation to the defect, and postoperative scars takes appreciable preoperative planning and discussion with the patient and family. The surgeon must match color, texture, and contour of the recipient site to the donor site to maximize aesthetic and functional outcome. The donor tissue must be free of infection, scars (or have stable scars), or trauma to prevent implant failure or extrusion. In most nevus cases an expander is placed through an incision within the border of the lesion. In other cases (such as

unstable scars, vascular tumors, and craniofacial deformities), incisions are planned outside the border of the defect or occasionally at a distant site.

The resulting skin flaps must be handled gently; rough or aggressive retraction or collision of the flaps with instruments can lead to skin-edge necrosis. The pocket is dissected to allow placement of the largest expander indicated, creating the pocket approximately 1 cm larger than the expander in all directions. Typically an internal remote port is used, with placement over a region of firm skeletal support (for example, the preauricular region, cranial vault, rib or iliac crest, or anterior thigh for trunk/abdominal TE) to ease outpatient filling. Partial fill of the expander (usually to 10% to 20% of its listed volume) ensures that the expander is properly positioned and without firm surface folds. Closed suction drains also serve to control potential dead space from wide undermining. In most cases the expander pocket incisions are closed in watertight fashion with judicious placement of 4-0 clear nylon buried intradermal sutures, followed by 4-0 Prolene running continuous suture. Skin flaps are dressed nonadherently (bacitracin, Xeroform gauze), followed by soft padding (fluffs).

Although the family receives thorough preoperative education about the tissue expander, parents are often more comfortable if their child remains in the hospital for observation for the first postoperative night. Patients may also be monitored overnight for potential compromise or hematoma formation and to manage pain control.

Serial injections are started 7 to 10 days after insertion of the expander, if the skin flaps are in excellent condition. After one or two postoperative visits for drain removal (postoperative days 3 to 10) and education (greatly aided by preoperative teaching), most pediatric patients are able to begin a home expansion protocol directed by parents or guardians. One study demonstrated that home expansion is as safe as office expansion, and the successful outcome rates are nearly equivalent (96% for office expansion versus 90% for home).¹⁴ Expansion should render the skin tense, but it should not be painful to the patient or compromise the skin. Either condition suggests excessive filling, which must be corrected immediately.

In our practice, families are given the option of home or office expansion. When the option for home expansion was introduced nearly 30 years ago, approximately 20% chose to expand at home. Today our practice manages nearly 80% of our patients with home expansion. Families typically proceed with a well-established expansion protocol for 8 to 12 weeks before the expander is removed and reconstruction can be performed. Parents record progress with digital images and transmit these by email correspondence during the expansion. The parents are also provided with a printed card to record the schedule and amount of saline solution injected throughout the expansion process.

The Internet has provided a powerful tool for following patients, even at a distance. It also gives families a measure of comfort, because they are never far from advice and support. It is important to obtain consent from parents and patients before communicating by email and using this for image transmission. Physicians should ensure the presence of data encryption systems and password-protected accounts with practice management, hospital administration, parents, and patients to preserve patient privacy and remain in compliance with the Health Insurance Portability and Accountability Act (HIPAA).

Although each case varies, depending on the underlying anatomy and reconstructive problem, a rough guideline for typical injection amounts by site is:

- Scalp: 20 to 50 cc
- Cheek: 5 to 10 cc
- Forehead or adjacent neck: 10 to 20 cc
- Trunk: 50 to 100 cc

TE in the extremities varies based on the size of the expander, the site on the extremity, and the age of the patient. Expanders are usually injected with saline solution once per week, and the overall amount is monitored closely so that midway through the process, the frequency of injections may be increased to every fourth or fifth day if it appears that the target expander removal date will be significantly delayed. If the desired volume is reached earlier than expected, an effort is made to move up the excision and reconstruction schedule.

Antibiotics

A broad-spectrum antibiotic is initiated when anesthesia is induced and is continued for 7 to 10 days postoperatively, depending on when the drain is removed. Previous studies support our own experience that ear infections and various viral infections may seed the expander site, resulting in an infection. By maintaining a low threshold for placing the patient back on a broad-spectrum antibiotic in the presence of an upper respiratory infection, ear drainage, or unexplained fever, most infections can be prevented or incipient infections squelched before the potential loss of the expander. Myringotomy and tube placement should be considered in patients with chronic ear infections before undertaking expander placement, balancing the risk of infection and expander loss against the risk of the myringotomy and tubes. The latter should especially be considered in a child who will undergo multiple rounds of expansion. With multiple expanders, infection leading to complications in one expander may be addressed without the need to remove other expanders. This is particularly important when the problem is encountered early in the course of expansion.

Expanded Flap Design

The design of an expanded flap is a significant undertaking. Although the early dogma of TE emphasized expansion as a means of generating large advancement flaps only, experience has demonstrated that expanded transposition and rotation flaps may frequently be preferable for many reconstructions.^{7,15} Clearly, the increased vascular supply of the expanded flap places little limitation on the ingenuity of the surgeon for designing flaps unique to the recipient defect. Although requiring more planning and forethought, transposition of the flap provides greater versatility in flap design and range.⁷ What differentiates an expanded transposition flap from an unexpanded one is that when the flap base is expanded, the flap not only transposes, but it also advances because of the tissue gained in the area of the pedicle. This allows significant movement of the flap (particularly in an axial direction) and therefore an improved ability to cover large surfaces with each expansion.

REGIONAL CONSIDERATIONS

We will discuss the considerations needed for successful pediatric TE in each body region based on nearly 35 years of experience with more than 3500 tissue expanders.

Scalp

There are three general indications for pediatric scalp reconstruction with TE: to surgically correct large congenital nevi, to correct scar and skin graft alopecia, and as an adjunct to craniofacial reconstruction.^{7,16} Despite previous concerns that expansion may affect cranial vault morphology, an early examination by CT analysis of more than 20 infants undergoing scalp expansion found no distortion or other untoward effects on cranial sutures.¹⁶ We have not seen a case of permanent

deformation of the skull in an expander patient in all our years of using TE. Although cranial molding may appear, this usually corrects itself within 1 to 3 months.

We delay placement of expanders in babies with significant positional plagiocephaly until the greater part of cranial remodeling has occurred. We typically place expanders for treating large and giant nevi at 6 months of age. Expansion for the treatment of extensive areas of cutis aplasia is typically delayed until the patient is at least 1 year of age, when the scar tissue is more stable.

Fig. 12-2 illustrates the use of TE with reconstruction for cutis aplasia. It also demonstrates that excessive scoring or excision of the thickened capsule in the area of the flap pedicle may result in possible flap compromise. Expanders are placed to allow maximal expansion of the areas of normal scalp that will provide the optimal hair orientation after the reconstruction, and the next round of expansion is always delayed at least 4 months. Expanders are never placed immediately at the time of nevus excision and flap repair. At subsequent rounds of expansion, a larger expander can be placed to distribute the expansile forces evenly over the hair follicles.¹⁶

As previous studies have shown, TE does not induce proliferation of hair follicles, but it can more than double the size of the scalp without a visible decrease in hair density. Fig. 12-3 demonstrates TE for the reconstruction of postirradiation alopecia. Expansion is effective but may be slow, and exposure of the expander because of poor healing may limit complete expansion but not compromise the final result. For expander placement, pocket dissection is performed subgaleally (and subfrontalis when combined with forehead expansion). Dissection is typically easiest with a graduated series of malleable retractors and pediatric urethral sounds to spread open the pocket with limited trauma to the overlying flap. Flaps are designed with consideration given to the major blood supplies to the scalp (that is, the superficial temporal, postauricular, occipital arteries, and contributions from the supraorbital vessels). Port placement in the preauricular region has produced the least migration.

Capsular tissue commonly creates a thick ridge of fibrous tissue along the cranium while the expander is in place. When the expander is removed and the flap advanced, the ridge is often visible and palpable, which can be distressing to the patient and parents. On removal of the expander, any remaining fibrous tissue will resorb and flatten, with the dissolution of the capsule in



Fig. 12-2 A and B, This 2-year-old girl had extensive cutis aplasia of the scalp.



Fig. 12-2, cont'd **C**, Three expanders were placed for 12 weeks of expansion (340 cc right, 250 cc left, and 70 cc occiput, with an external port for the occipital expander because of the limited placement site). **D**, During resection, an overzealous backcut of a thick capsule resulted in partial loss of the right-sided flap. A second expansion was performed 1 year later, resulting in excellent coverage. **E-G**, The patient is seen 6 years after reconstruction.

6 to 8 weeks. Although it may be tempting to excise this tissue to smooth the surface, it should be stressed that the capsular ridge contains a valuable vascular supply to the flap. Along the pedicle or trailing side of the flap, the capsular ridge should not be excised; doing so will lead to flap compromise. Similarly, we advocate against scoring of the capsule to gain extra length, because this will also have a negative impact on the vascularity and venous drainage. On the leading edge of the flap to be transposed, the capsular ridge can be sharply dissected from the skull to aid in flap inset and advancement.

Second to the breast, the scalp is likely the most frequently expanded region and the area with which most surgeons are familiar. The ability to reconstruct areas of scar alopecia, resect and reconstruct large scalp lesions, and replace defects with hair-bearing scalp makes expansion the reconstructive option of choice for many patients.

One technique that has proved valuable for complex reconstructive cases, particularly with scar alopecia (but equally valuable for scars, wounds, or defects at sites other than the scalp) when





Fig. 12-3, cont'd H-J, Fifteen months after surgery.

there is minimal available donor tissue is “in situ” serial expansion. If serial expansion is required to fully reconstruct a defect, yet the scar tissue is too tenuous to allow adequate healing (after the first flap advancement) for performing subsequent expansions without risk of expander extrusion, the initial expander can be fully expanded and then switched for a larger expander in the same pocket. The flap can then be further expanded before the first flap advancement is performed. This swap of expanders can be carried out through a minimal incision, and the new expander can be connected to the same injection port. Generally, the second expander can be expanded to the same volume as the one being replaced at the time of the switch. Because a significantly larger flap has been created, it can safely move beyond the region of unstable scar.

The use of expanded transposition flaps rather than simple advancement flaps has greatly reduced the number of serial expansions required and has resulted in improved reconstruction of hair direction and hairline (particularly the temporal hairline). Fig. 12-4 illustrates expansion of an occipital transposition flap and hemiforehead flap for reconstruction of the parietal scalp, temporal hairline, and forehead in a patient with a giant nevus. In hemiscalp or near-hemiscalp reconstruction for a nevus or scar alopecia, temporal hairline reconstruction is best accomplished with a large transposition flap from the occipital region.⁷ In the young boy with a large facial nevus shown in Fig. 12-5, the design and staging of forehead, nasal, cheek, and scalp reconstruction are demonstrated, with staged forehead, scalp, and neck flaps.

Forehead

Expanded flap reconstruction of the forehead provides some of the most challenging cases because of its potential morbidity and its potential to disfigure the upper facial structures such as the brow. The surgeon must respect the aesthetic subunit to avoid late complications. A retrospective review of our early cases revealed a 24% aesthetic complication rate with forehead TE, including brow asymmetry, brow ptosis, altered hair direction, and anterior hairline asymmetry¹⁵ (see Figs. 12-4 and 12-5).



Fig. 12-4 A and B, The patient is shown at initial presentation. C and D, Starting when he was 6 months of age, two 250 cc expanders were placed under the normal forehead skin and the occipital scalp behind the nevus. E, Before first-stage partial excision of the nevus and removal of expanders. The buried port can be seen below the nevus. F and G, Shown immediately postoperatively, following the tenet: “Don’t overextend—reexpand.”



Fig. 12-4, cont'd H and I, Four months after the first stage. J, In the second round of expansion, two 250 cc expanders were placed under the previously expanded forehead and parietal scalp. K and L, The patient is shown 1 week after the second stage with completion of the excision and expander removal. M, The first day of preschool, 18 months after scar revision for the brow.



Fig. 12-5 This patient presented with a large facial nevus. Staged forehead, scalp, and neck flaps were planned. **A**, When he was 6 months of age, a 250 cc expander was placed in the hemiforehead and under the occipital scalp for 12 weeks of expansion. **B** and **C**, Four months after first-stage reconstruction of the nose and part of the forehead, the temporal hairline has been put into correct position, abutting the facial nevus that will be resected in the coming surgeries. **D**, The second round of expansion was started 6 months after the first resection and focused on the remaining forehead and the cheek. A 250 cc expander was placed under the previously expanded forehead flap, and a 70 cc expander was placed in the lower cheek to reconstruct the cheek and lower temporal hairline; expansion continued over 10 weeks. (The cheek can also be done with a 250 cc expander in the neck of a child, depending on the size of the area needing reconstruction.) **E** and **F**, Four months after second-stage reconstruction of the forehead and cheek. **G**, Third and final expansion of the forehead flap with another 250 cc expander, which was placed 6 months after the previous surgery and expanded for 10 weeks. **H**, Immediately after completion, excision of the forehead nevus.



Fig. 12-5, cont'd I, Four months after the final expander stage. J-L, The boy is seen 18 months after final expansion at age 3½. A scar revision of the brow had been performed in the interim.

Most of these complications are corrected with minor secondary procedures, but over the years certain guiding principles have been developed to minimize these late complications:

- Bilateral expansion of normal forehead tissue is often successful for midforehead lesions. Although completion of the aesthetic single unit forehead reconstruction should be considered if there is a wider area of normal forehead skin available to expand for a central defect.
- As with the scalp, serial expansion of the forehead is often required for hemiforehead reconstruction. The surgeon should remember: “Don’t overextend the flap; reexpand it.”
- Supraorbital and temporal lesions and scars are managed with a combined advancement and transposition of expanded normal skin medial to the lesion or defect.
- With minimal involvement of the temporal region, parietal expanded scalp can be advanced to reconstitute the hairline.
- When advancing and transposing the forehead flap, it is necessary to set the position of the brows as a primary step. We use three or four judiciously placed 4-0 clear nylon sutures to tack the deep brow tissue to the periosteum along the superior orbital rim. Paying close attention to obtaining symmetry will help reduce the need for secondary procedures. This also helps to prevent distortion of the contralateral brow.

- It is easier to elevate a brow later than to bring an elevated brow down. In cases of brow elevation, the abnormal brow can be returned to its preoperative position by interposing non-hair-bearing forehead skin. However, it does not work to simply expand the adjacent forehead skin to reorient the brow, because this tissue will recoil postoperatively. There must be some transposition of the tissues and resuspension of the brow itself to prevent relapse of the defect.

Opinions vary regarding the efficacy of expanding forehead flaps for nasal reconstruction with complex congenital nasal defects and acquired loss of nasal tissues. Although the donor wound on the forehead can be left to contract quite successfully in adults, a wide paramedian scar can be particularly disfiguring in a child. This scar can be minimized with additional tissue gained by TE (Fig. 12-6; see Fig. 12-5). An expanded forehead can also be used for upper eyelid reconstruction by transposing a flap inferior to the eyebrow. Although the pretarsal eyelid is best reconstructed with a skin graft, the preorbital eyelid tissue is often thicker and benefits from the thin layer of subcutaneous fat that is brought with an expanded forehead flap in this area (Fig. 12-7).



Fig. 12-6 Although opinions vary on the need for expansion of a forehead flap for nasal reconstruction, expansion allows tension-free closure of the donor site in a child. **A** and **B**, This child was born with total agenesis of the right side of the nose and hypoplasia of all nasal structures (including minimal choana on the left, which was associated with a midline cleft lip and multiple other congenital anomalies). She is shown in infancy before any facial surgery and at 3 years of age after an initial attempt at reconstruction without adequate skeletal support or reconstruction of the nasal airway.



Fig. 12-6, cont'd C-E, When she was 5 years of age, a 250 cc expander was used to expand the forehead, and first-stage reconstruction was performed on the left nasal airway with bone resection and flap lining. F and G, Expanded flap reconstruction over combined rib and cartilage grafts with completion of the left nasal airway reconstruction and creation of a false nostril on the right side. H-J, The result is seen 11 years postoperatively when the patient was age 16. She had not undergone any further surgery.



Fig. 12-7 Reconstruction of the upper eyelid and nose can be completed with expanded forehead skin for an optimal color and texture match. **A**, This patient presented at 6 months of age with an isolated, complex nevus of the left upper lid, brow, and nasojugal area. **B**, A 250 cc expander was placed in the ipsilateral forehead and was expanded for 8 weeks. **C**, The child is shown 8 months after expanded forehead flap reconstruction of the upper eyelid and nasojugal region with an island scalp flap for the eyebrow. **D**, The patient is seen at 2 years of age, 3 months after scar revision and contouring of the flaps around the canthal region, plus microhair grafting for the brow. **E**, The boy is seen at 5 years of age without further surgery.

Face and Neck

To achieve an optimal aesthetic and functional result in the facial and cervical regions, the surgeon must adhere to a subunit principle.¹⁷ This principle dictates that incisions be placed so that the scar is hidden in a natural crease, such as the nasolabial fold. Analogous to upper facial structures (such as the brow and the eyelid), undue tension on the lower face can disfigure critical structures, leading to lower lid ectropion, lip drooping, and oral incompetence. Advancement of cervical skin flaps cephalad to the cervicomandibular angle bears an increased risk of late complications. Neale et al¹⁸ reported a 10% lower rate of eyelid ectropion and a less than 10% lower rate of lip deformity in this context. The use of expanded transposition and rotation flaps from the lateral cheek, neck, and postauricular area minimizes the risk of these problems.^{7,15} Early in our practice, we based neck and cheek flaps medially, with the transposition occurring along the preauricular region and the zygoma, as a Mustardé-type lateral cheek flap. However, this design required frequent revision for ectropion because of the lack of supporting tissue for the flap. In addition, this design often required repeat expansion to complete the aesthetic unit reconstruc-

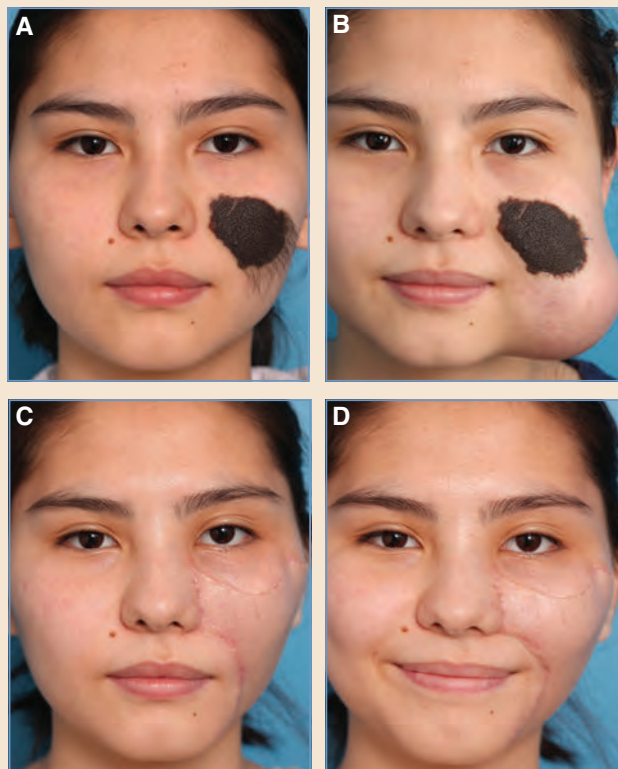


Fig. 12-8 A, This patient was 16 years old when she presented to our practice with an intermediate-sized congenital pigmented nevus. B, A 70 cc expander was placed in the cheek adjacent to the nevus, and she underwent 8 weeks of expansion. C and D, The girl is seen 1 month after surgery, with scars settled nicely along the borders of facial subunits, especially along the nasolabial and mental creases.

tion. In our current practice, the cheek, lower lid, and nasojugal lesions are treated with an expanded flap based laterally; transposition takes place from the submental neck, with incision lines placed along the nasojugal, nasolabial, and mental creases (Fig 12-8.) Over time, we have noted that by securing the expanded capsule along the periosteum of the piriform process and the medial and lateral orbit, the flaps are well suspended. Fig. 12-9 shows the use of the capsule to aid in suspension of the flap by anchoring it to the periosteum of the orbital rim and piriform process. Judicious flap design, the use of multiple expanders, and overexpansion are recommended to further minimize complications.¹⁸

Expanded full-thickness skin grafts can also achieve better functional and aesthetic results than split-thickness grafts; using the benefits of expansion will virtually eliminate graft size as a limitation for choosing this reconstructive modality.¹⁹ Preexpansion of donor sites allows larger full-thickness skin grafts for areas such as the periorbital or dorsal aspects of the hands and feet. Part of the expansion provides for the graft tissue or free flap, with the remainder providing tissue for primary closure of the donor site. The neck above the clavicle is the ideal donor site for grafts to be placed on the face (for example, a single expanded full-thickness skin graft to the entire forehead subunit) because of the excellent color and texture match. These expanded full-thickness grafts have all the characteristics of their unexpanded counterparts with regard to

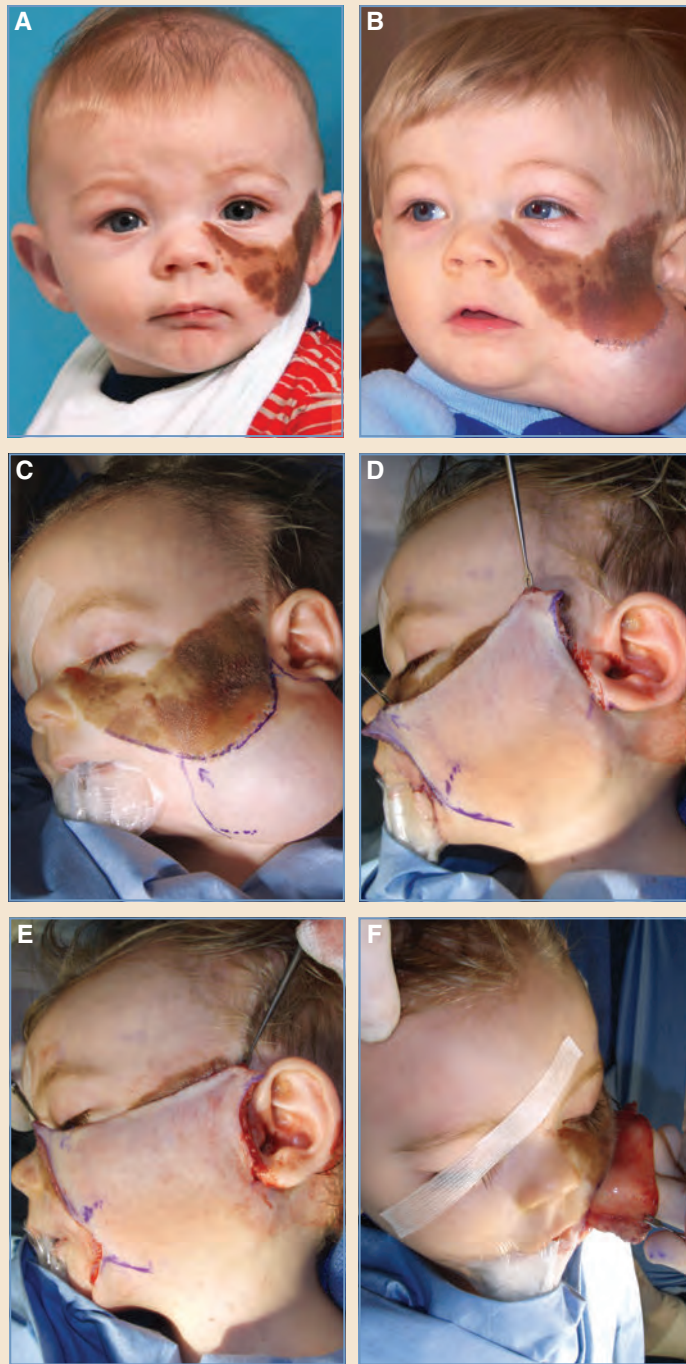


Fig. 12-9 A, This patient was 1 year old when he presented with a variegated, large congenital pigmented nevus of the entire cheek subunit. B, A 70 cc expander was placed into the lower cheek and neck, and he underwent 10 weeks of serial expansion. C-E, Operative steps show the incisional planning. By placing sequential incisions into the flap at the point where the nasolabial crease meets the mental crease, the medial aspect of this flap transposes into the nasojugal region and extends the tip toward the medial orbit for suspension. The capsule is shown extending all the way to the edge of the flap. This will be used to secure the flap to the periosteum.

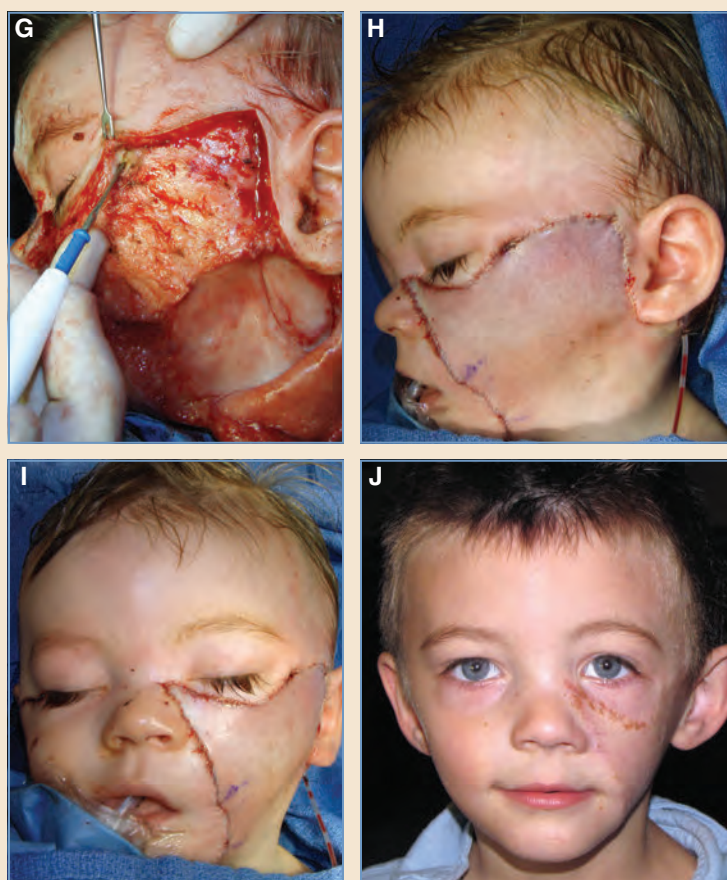


Fig. 12-9, cont'd G-I, Using a bovie or sharp scissors, a small area of periosteum is exposed at the key anchor sites, and 4-0 clear nylon sutures are placed to suspend the flap. J, The patient is seen 3 years after surgery at 4 years of age. The flap is well suspended without ectropion.

durability, texture, minimal contraction, and growth. Fig. 12-10 illustrates the use of expansion to allow harvest of a full-thickness skin graft to cover the eyelids and periorbital area in a single unit from an expanded supraclavicular donor site. It also demonstrates the use of a portion of the expanded forehead flap to cover the nasal dorsum after nevus excision. Although in the past we used a single-piece expanded full-thickness skin graft for coverage of the periorbital region (including both eyelids), today we prefer to use one of two postauricular grafts with a full-thickness skin graft from the groin to the postauricular donor site.

Preexpansion is also useful for distant pedicled flaps (such as the supraclavicular artery flap to the lower face) and free tissue transfer for similar reasons as discussed earlier. The delay principle can enlarge the angiosome territory that can be captured on a single pedicle. Free flap donor-site morbidity is also an important consideration. Grafting of donor sites suffers the same color and contour mismatch as if one were grafting a primary defect. Although these sites are often on the trunk or lower limbs, they are no less important when it comes to considering the lifelong impact that such a deformity can have on an individual's functional status and self-image. Free flap harvest from distant sites can reconstruct complete cheek or forehead aesthetic units when regional tissue is not available to expand for flap tissue. Fig. 12-11 illustrates expansion of a distant flap for





Fig. 12-11 A and B, This child was born with a congenital giant pigmented nevus involving the entire forehead, midfrontal scalp, brows, and nasal dorsum. Biopsies at 5 months of age demonstrated infiltration of the nevus through the underlying frontalis muscle, contraindicating expanded full-thickness coverage of the muscle and forehead aesthetic unit. When she was 18 months of age, the forehead nevus was excised after expansion of the adjacent normal scalp, leaving the forehead, brows, and nose. C, At 2½ years of age a 500 cc expander was placed in the right back, adjacent to a planned free scapular-parascapular flap to allow primary closure of the donor site, with a delay carried out 1 week before transfer. D and E, The flap was elevated and transferred with a vein graft to the superficial temporal artery, allowing excision of the nevus, except for a segment left for each eyebrow. F, One week after inset of the flap and before subsequent contouring of the nasal area. G, Six months postoperatively. H-J, The patient is seen 9 months after resection of the remaining nasal nevus and contouring of the flap.

free tissue transfer to reconstruct the entire forehead aesthetic unit and a portion of the nasal dorsum. Free flaps are also valuable for large defects involving the extremities (discussed later). In all cases, the expansion is carried out adjacent to the planned free flap and away from the pedicle.

Trunk

Beyond its obvious usefulness for treating deformities of the breast, TE has multiple applications on the trunk for treating giant nevi, vascular malformations, and contour defects.²⁰ It is also a useful adjunct to other procedures for closing congenital or acquired chest, abdominal, and back defects. Not unexpectedly, the lower abdomen is perhaps the most easily expanded site and can be used when excising adjacent lesions, as donor tissue for expanded pedicled flaps or flaps for free tissue transfer, such as for expanded transverse rectus abdominis myocutaneous (TRAM) flaps, where expansion is carried out to aid in donor-site closure, or expanded upper abdominal flaps to help close a large deep inferior epigastric perforator flap defect (Fig. 12-12), and as a donor site



Fig. 12-12 A combined regional expansion on the extremity and expansion of the TRAM flap for microvascular transfer. **A**, The patient presented at 8 years of age with a mixed lymphaticovenous malformation of the thigh and lateral knee associated with chronic drainage, pain, and limitation of activity. The lesion was too extensive to allow excision and reconstruction with regional expansion alone. However, regional expansion was planned to reduce the overall size of the expanded free TRAM flap that was planned for reconstruction of the greater portion of the defect. **B**, Expanders of 500 cc each were placed at both sites and expanded over 11 weeks. The abdominal expander was placed adjacent to the flap to allow harvest of a large flap with tension-free closure of the donor site. **C-E**, Resection and repair with advancement of the adjacent expanded flap and inset of the free TRAM flap.

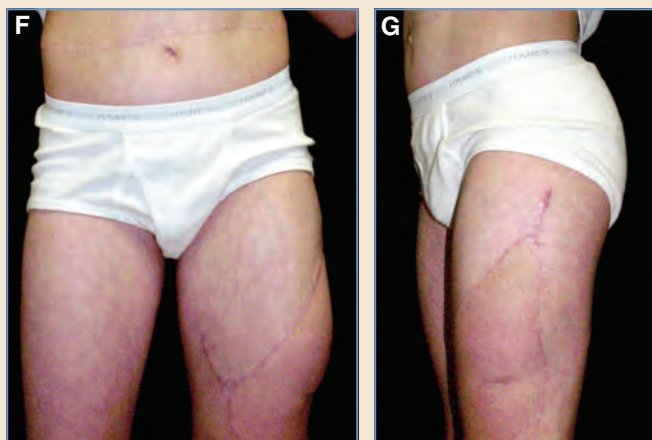


Fig. 12-12, cont'd F and G, The patient is seen 3 years after resection and reconstruction, before minor contouring of the flap with liposuction.

for large expanded full-thickness grafts.¹⁹ Expansion on the anterior trunk is limited in females by the need to avoid potential distortion of the breasts. For males, temporary movement of the nipple-areola complex can be corrected at the completion of the flap reconstruction by grafting it back in the appropriate location. For the trunk, keeping in mind the need to place the injection port over firm skeletal structures, the injection ports are frequently placed in the anterior thigh or, less frequently, over the anterior ribs.

Expansion on the posterior trunk has become the modality of choice for treating many large and giant nevi of the back and buttock area. Whether advancing skin caudally or cephalad, serial expansion is frequently required to excise extensive lesions. Expansion can begin as early as 6 months of age for treating large and giant nevi in children and is clearly easier in the earlier years than later in life.

For patients with lesions that are circumferential or that involve both an anterior and posterior component, we stage the expansions, preferring to begin on the back and then placing front expanders in a second round or third round. Typically, for a large nevus on the back, flanks, and abdomen, two expanders are placed in the upper back/chest/flank, wrapping down along the upper margin of the nevus and positioned just off midline medially. When expanded, these flaps provide large transposition flaps and the largest degree of flap movement. These are then expanded in the next round to move further caudal as well as laterally. Today it is not uncommon to have expanders in place on both the anterior and posterior trunk.

The lower back may be expanded to develop large transposition flaps to cover the buttock, and the lower abdomen can be expanded to create similar flaps for coverage of the anterior thigh. Again, the use of large expanded transposition flaps has allowed excision and reconstruction of giant nevi with fewer procedures and more aesthetic and functional positioning of the final scars^{7,21} (Fig. 12-13). The same principle can be carried out by moving the tissue from an uninvolved hemiabdomen across the abdomen and lower chest when the normal tissue is unilaterally placed and the normal tissue in the lower chest cannot be expanded without risk of distorting the breast. Expansion of the lateral abdomen, flank, and/or back is also used commonly to develop large expanded pedicled flaps for reconstruction of large nevi on the upper extremity.



Fig. 12-13 This young child underwent early excision of most of his giant nevus at 2 months of age and reconstruction with a thin-meshed, split-thickness skin graft and Integra. However, graft take was poor, and there was severe hypertrophic scarring. TE was used as a means of secondary reconstruction for treating the scar deformity. **A**, At the time of his presentation at 2 years of age, the patient had significant limitation of movement, severe pruritus, and pain associated with the scarring. Staged expansion of the back and shoulders was planned to excise the graft and scar tissue. **B**, Expansion of the two 750 cc expanders in the lower back and the 250 cc expander in the right shoulder is nearly complete. They were placed at 2½ years of age and expanded over 11 weeks. **C**, The result is seen immediately after initial partial excision and flap reconstruction. **D**, Two months postoperatively. **E**, At 3½ years of age, 6 months after removal of a second set of expanders and further resection of graft and scar, and before placement of a third set of expanders. **F**, Early in the third serial expansion. Two 750 cc expanders were used in the upper right and lower left back, and a 250 cc expander was used in the left shoulder.



Fig. 12-13, cont'd **G**, After 10 weeks of expansion. **H**, Result 5 months after completion of the excision and reconstruction. **I** and **J**, The patient is seen at age 5, 1 year after his final surgery.

Expansion has been used as an adjunct for treating secondary problems associated with myelomeningocele more commonly than for early repair. Expanded cutaneous and fasciocutaneous flaps can provide more stable coverage for planned spinal fusion and for salvaging exposed hardware when complications arise.

Congenital Breast Anomalies

Expanders and expandable implants have found roles in postmastectomy breast reconstruction and habilitation of congenital breast anomalies.²⁰ Much of this is covered in Chapter 60, but a few comments seem pertinent here with regard to reconstruction for congenital breast anomalies.

Expanders can be helpful as “spacers” when correcting congenital breast anomalies. Requests for breast habilitation come from patients with breast agenesis associated with Poland’s syndrome, idiopathic unilateral breast hypoplasia, and iatrogenic breast asymmetry caused by juvenile

breast bud damage. Traditional wisdom has been to wait for maturity before correcting breast asymmetries. This strategy ensures that the surgeon knows what needs to be matched on the opposite side. Although this solution may have been acceptable at one time, today's young adolescent female has problems with changing in locker rooms, participating in sports activities, and wearing fashionable clothing. Questions of developing self-esteem, body image, and sexual identity further compound the issue of waiting. Expanders can function as an intermediate solution.

Many young women are happy with breast volume symmetry so that they appear normal in a bra. With this goal in mind, an expander can be placed as early as the opposite breast begins to develop and can be expanded over time. Aesthetic surgeons also use this method to adjust breast size over time.²⁰ The use of either an expander or an expandable implant in these patients may constitute an off-label use for these devices. Expanders are not constructed for prolonged use (that is, over the 6 to 8 years of breast development). Adjustable permanent implants are designed for longer-term use but are supposed to have their ports removed promptly after complete fill to prevent implant deflation. A discussion of these possibilities needs to occur with the patient and caregivers before implant placement. In general, it has not proved to be a significant problem in practice.

The postoperatively adjustable implants come in sizes, shapes, and volumes particularly suited to this situation. Depending on how much native breast exists, these expanders can be placed subglandularly or submuscularly.

After the patient reaches maturity, the expander can be replaced with a permanent implant, and balancing procedures to match breast shape can be performed on the opposite breast for "out of a bra" symmetry. In the patient with Poland's syndrome with significant infraclavicular soft tissue deficiency, a latissimus dorsi flap can be transferred or a custom permanent implant can be manufactured at the time of definitive reconstruction.

Extremities

Classically, the extremities have been viewed as sites where TE is of limited value and is associated with a higher risk of complications. Complication rates in the limbs have been shown to be significantly higher than non-limb sites (47% versus 23%) by Casanova et al.²² They also found an overall 19.4% complication rate in their 10-year retrospective review of more than 200 cases, including a 15.5% major complication rate and a 4.9% failure rate. Despite these prevailing opinions and ominous reports, expansion in the limbs is feasible.^{21,23} The surgeon must carefully assess the lesion or defect to be treated and understand the limitations inherent in the basic geometry of the extremities (and understand that flap movement is easier around the circumference of the limb than axially). Careful planning, choice of expander size, placement through remote incisions (in selected cases), and a possibly slower expansion schedule will lead to a lower risk of infection, extrusion, and flap failure. Unstable infected wounds are relative contraindications to TE in an extremity. Expanded pedicled flaps and expanded free flaps, such as the TRAM, deep inferior epigastric perforator (DIEP) and scapular flaps, are also powerful options for reconstructing large extremity defects.²³

For upper limb reconstruction we have devised a useful algorithm for complex defects.²³ In our experience, obtaining successful contour and color match of the upper extremity comes from approaching it in thirds (proximal arm to elbow, forearm, and the hands) and noting whether the lesion is circumferential or noncircumferential. For proximal noncircumferential defects, expanded transposition flaps from the back or shoulder serve well. If the lesion is large and circumferential, covering most of the proximal arm, and the nevus involves the adjacent back or shoulder skin, an expanded free flap is the modality of choice. Distally, for large circumferential forearm lesions, expansion of the flank creates a large pedicled flap through which the forearm can be placed during vascularization of the flap from the recipient bed (Fig. 12-14). After 3 weeks,



Fig. 12-14 Excision and reconstruction of a circumferential nevus of the arm with an expanded pedicled flap from the abdomen and flank. **A**, The patient is seen at 6 months of age, with a nevus extending from above the elbow to the wrist. **B**, The 750 cc tissue expander is shown after 12 weeks of expansion (note that the expansion is carried up onto the flap and back). **C** and **D**, The flap before division of the pedicle and after excision of all but a thin border of proximal and distal nevus. Sutures were placed to narrow the pedicle and ensure that the expanded skin gained will both fully wrap the arm and allow primary closure of the donor site. **E** and **F**, The flap was divided after 3 weeks of attachment.

Continued



Fig. 12-14, cont'd G and H, Immediately after inset of the flap. I and J, Result after excision of the remaining nevus, which was carried out 8 months after the initial flap inset. K and L, Appearance of the flap and donor site when the patient was 4 years of age, 2½ years after flap reconstruction.

the pedicle is divided. Early placement of “crimping sutures” through the pedicle helps to ensure the correct flap size at division and inset and adequate tissue for closing the donor site. Given the broad area of attachment for these flaps, complex immobilization is not required if the flap has been carefully planned so that the extremity is in a comfortable position of rest during attachment.

As previously reported, expanded full-thickness skin grafts from the abdomen or groin remain the treatment of choice for covering extensive nevi on the hand.¹⁹ Regional expansion can be used for select smaller lesions and scars of the arm and dorsum of the hand when a donor area large enough for placing the expander is available.

Expansion, although useful for treating intermediate-sized lesions of the lower extremity, is even more limited in its applications there than in the upper extremity. However, with experience, a surgeon can use expansion safely with limited risk of infection or extrusion. Expansion is easier above the knee than below it. Because of the geometry of the extremity, flaps move more easily around the circumference of the limb than axially (Fig. 12-15). Below-knee movement, even transversely, is limited. When possible, more than one expander should be used at a time. Likewise, ports should be kept at distant sites, and the expanders should be placed through smaller, nondependent incisions as much as possible (Fig. 12-16).



Fig. 12-15 TE in the thigh is limited by the geometry of the extremity and restriction of flap movement. This case of a large nevus demonstrates the need to use more than one expander when possible and the increased movement of the expanded flap that can be gained with a transposition flap versus an advancement flap. **A**, The nevus is shown before surgery in this 8-year-old child. **B** and **C**, The 750 cc and 250 cc expanders are shown after 12 weeks of expansion.

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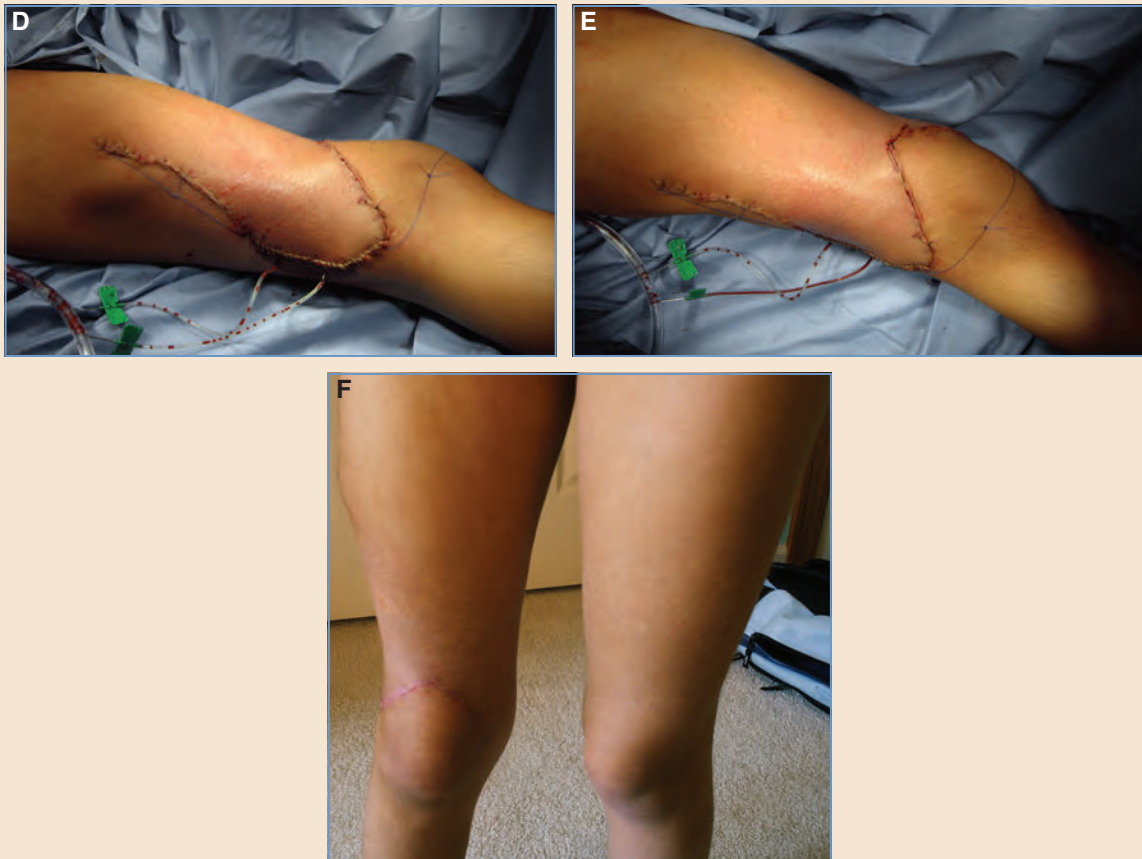


Fig. 12-15, cont'd **D** and **E**, The nevus was partially excised, leaving only a small rim of nevus in the area of the drains lateral to the knee. Transposition of the flap allowed optimal placement of the scars and avoided distortion of the thigh contour. **F**, The result is seen 1 year after excision of the final portion of the nevus, which was carried out 6 months after the initial excision.



Fig. 12-16 Soft tissue reconstruction of the lower extremity after necrotizing fasciitis. **A and B**, The patient presented at age 2 with a necrotizing infection that was treated with serial debridement and a meshed split-thickness skin graft. **C**, Nearly a year later, a 250 cc expander was placed in the proximal adjacent skin of the lower leg, and a 70 cc expander was placed distally near the ankle. **D**, After 11 weeks of expansion, part of the skin graft was excised, and the limb was reconstructed with expanded flaps. **E-G**, Four months after first-stage reconstruction.

Continued



Fig. 12-16, cont'd H and I, A second round of expansion was done 11 months later. This time a 500 cc expander was placed below the proximal flap, and a 70 cc expander was placed beneath the ankle flap. J and K, A short time after the second round of expansion and reconstruction after 11 weeks of expansion. L and M, Six months after the second set of surgeries, another 500 cc expander was placed into the leg. N-Q, The patient is seen at 6 years of age, 1 year after final expansion.

Treatment of large extremity defects may be better accomplished by using expansion as an adjunct to reduce the overall size of the defect, which can then be covered by free tissue transfer (the skin adjacent to the donor site is frequently expanded to allow harvest of larger flaps with primary closure of the donor site). In select cases, TE of pedicled flaps from the buttock and thigh may be used to cover the extremity below the knee after resection of a giant nevus, although this type of flap needs to be carried out in children under 1 year of age. Children older than this typically lack the flexibility to bring the lower leg against the thigh and buttock for the necessary 3 weeks of flap attachment (Fig. 12-17).



Fig. 12-17 An innovative way of using an expanded pedicled flap to cover a circumferential nevus from knee to ankle, an area where TE is of limited use, and the long-term result of grafting is often poor. **A and B**, This 1-month-old infant had a nevus with marked variegation of color, texture, and surface characteristics. The flexibility of the leg and position in relation to the planned posterior thigh donor site is demonstrated. **C**, The 350 cc expander, which was placed through an incision in the buttock region when the patient was 2 months of age, is shown near completion of expansion.

Continued



Fig. 12-17, cont'd D and E, After an intermediate delay of the flap to augment the medially based axial vascular supply, the nevus is excised and the flap wrapped around the leg, slipping the foot through the bipedicle flap that was created between the expander placement incision and the incision at the superior border of the flap. This bipedicle flap was used as the only immobilization of the flap, in addition to its large area of expanded flap attachment. F, Three weeks after nevus resection and flap attachment, before flap division and inset. G, The immediate postoperative result showing flap inset and use of the bipedicle flap, now converted to a caudally based unipedicle flap to augment tissue available for donor-site closure of the thigh.

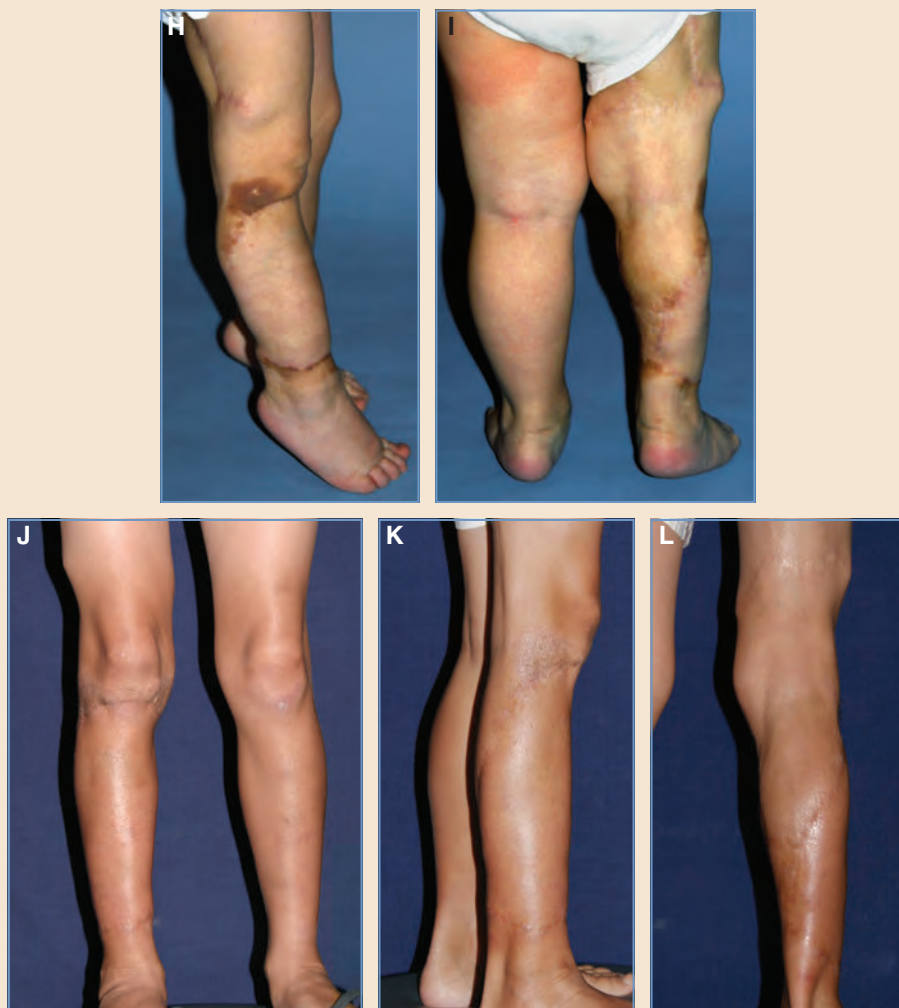
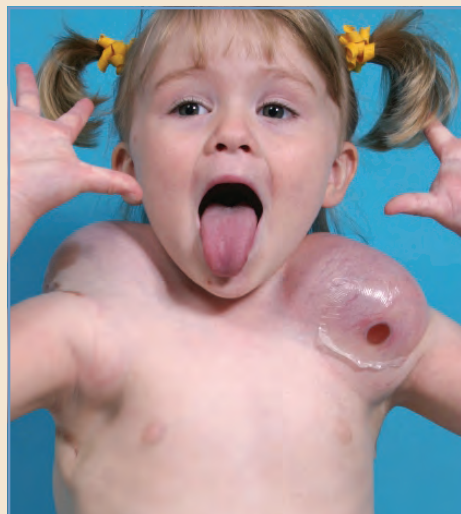


Fig. 12-17, cont'd H and I, The result is shown 1 year after excision and expanded flap reconstruction. J-L, The patient is seen at 7 years of age, 4 months after further excision of the nevus along the scar margins.

MANAGEMENT OF COMPLICATIONS

Beyond the site-specific complications of TE mentioned previously, general complications of expansion are divided into major and minor categories. Major complications may involve infection, implant exposure, extrusion, and flap ischemia.^{15,24} Traditionally it has been taught that early postoperative infections should be managed with expander removal and antibiotics; however, as noted previously, careful attention to subtle signs of infection and maintaining a low threshold for placing the child on antibiotics again often will often prevent loss of an expander. We have also found that if a child is developing fevers and there appears to be fluid around the expander, this can be safely drained from the remote port site. This often relieves the fever, and with the use of antibiotics, can salvage the expander. The surgeon should also recognize that the augmented

Fig. 12-18 Placement of a semipermeable transparent film over exposed expanders helps protect the flap and secure the open site, preventing extrusion. Often this dressing will stabilize the wound enough that when multiple expanders are in place, the patient can complete the expansion regimen for the other expanders while maintaining the expansion already gained in the exposed expander. Reconstruction can then be carried out without loss of tissue gain.



vascular supply of an expanded flap allows safe movement of the flap to its full extent when removing an infected or exposed expander, because the primary source of the problem (the foreign body) is being removed. Careful wound irrigation with an antibiotic solution and wound drainage provide additional margins of safety.

Extrusions can be dealt with similarly. If a small area of the incision breaks down adjacent to the expander, yet the surrounding wound is stable, the expander may be salvaged and some additional expansion gained (reducing the amount injected at each expansion and varying the injection frequency). We have found that an expander is more likely to be salvageable if it lies in a dependent position in relation to the open wound. Such extrusions are also managed conservatively with local wound care and oral antibiotics. In our practice the use of semipermeable transparent dressings (Opsite, Tegaderm) to cover an exposed expander helps to support the surrounding skin and can often aid in salvage when applied in a timely manner (Fig. 12-18). Under such conditions some wounds will remain stable, with only a small area of the expander exposed, allowing completion of the intended expansion.

There are a number of minor complications associated with expansion. Pain during expansion is typically transient; however, pain noted during a fill that persists can be a sign of overexpansion. Assessment of the flap with capillary refill and firmness as a guide will alert the surgeon to this problem. Removal of some fluid will protect the flap and usually treat the pain. New-onset pain that is not temporally related to the weekly fill can often be an early sign of infection, and this should be addressed as soon as possible. Other complications include seroma, dog-ears at the donor site, partial separation at the junction points of multiple incisions, minor distal flap ischemia, and the widening of scars. Collectively, these minor events occurred at a rate as high as 17% in one study.²⁴

CONCLUSION

Tissue expansion is a powerful reconstructive tool for the treatment of a broad spectrum of pediatric congenital and acquired deformities. The augmented vascular supply gained during the expansion process allows considerable flexibility in flap design, often limited only by a surgeon's ingenuity. Successful TE in children and adults begins long before placement of the tissue expander. Careful case selection and planning are critical, as is a clear understanding of the potential limitations and complications associated with expander use.

Parents and children (when old enough) must understand why expansion is being used, the time commitments necessary for completion of the expansion, and the limitations placed on activity during the process. Preoperative teaching by an experienced nurse allows parental involvement in the expansion process, provides the groundwork for them to vigilantly observe skin changes, and acts as an avenue for rapid communication of any concerns. Combining all of these considerations, TE is extremely well tolerated by children of all ages, and even in the face of occasional complicating factors, it provides a successful approach for treating many reconstructive problems.

KEY POINTS

- Successful expansion, regardless of indication, relies on careful case selection.
- TE in children is not associated with a higher risk of complications than in adults, and it is extremely well tolerated.
- Safe expansion in the pediatric population begins with careful assessment of each patient. This includes determining whether TE is the optimal technique for the reconstruction planned, whether the patient and/or the parents will be able to manage the routines necessary, whether there is a clear understanding of the potential complications, and whether the time constraints inherent in the process can be accommodated in the specific situation.
- When appropriate, TE at home is possible, safe, and advantageous to the patient and family. It requires close interaction and preoperative and postoperative instruction by the nursing staff and physician.
- The surgeon must understand the potential problems and avoid them.
- The choice of expander, incision for placement, and expansion routine may vary according to the site of expansion and the underlying defect being treated.

Continued

KEY POINTS (continued)

- TE can safely begin in the scalp at 6 months of age, and some cranial molding is to be expected, but there has been no demonstrated risk of long-term cranial deformation.
- Expanded transposition flaps in the head and neck, trunk, and, to a lesser extent, the extremities allow much greater use of expanded skin flaps than traditional expanded advancement flaps.
- The average length of time between placing an expander and reconstruction with flap movement is 10 to 11 weeks, with 3 to 4 months between each set of expanders for serial expansion.
- The combined use of expanded flaps and expanded full-thickness skin grafts provides optimal aesthetic and functional reconstruction in the face and periorbital area, with the former used for reconstructing the forehead and cheek and the latter used for the periorbital area and eyelids.
- Our experience has demonstrated that a higher rate of complications in nonnevus cases may require variation in technique.
- Expansion can be used safely in the extremities but is limited, because expanded flaps move better for coverage circumferentially than axially.
- Expanded pedicled flaps from the abdomen and flank can be used effectively to cover large circumferential defects of the upper extremities in infants and children.
- Perhaps the most important guiding principle for avoiding complications (especially flap compromise) is to reexpand flaps rather than try to overextend them.

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13

Radiologic Management of Vascular Anomalies

Philip R. John



There are two broad categories of vascular anomalies: vascular tumors and vascular malformations. This original division, which was proposed by Mulliken and Glowacki,¹ was based on the biologic and clinical behavior of these anomalies, including their physical characteristics, natural history, and cellular features. Since then, additional disease entities of varying complexity have been identified, some of which are not easily defined by the original classification. An updated classification was presented in 2014 at the Twentieth Congress of the International Society for the Study of Vascular Anomalies (ISSVA) and is now accepted as the most up-to-date classification of vascular anomalies.² This updated ISSVA classification has modifications with expansion of the subcategories² (Boxes 13-1 and 13-2). This more comprehensive classification provides an excellent framework for the medical community to use when managing patients with vascular anomalies.

A few vascular anomalies remain unclassified because of uncertainty whether they are tumors or malformations, or because their clinicopathologic features are not fully understood. Included in this group are Verrucous hemangiomas and angiokeratomas, kaposiform lymphangiomatosis, and PTEN hamartoma of soft tissue.²

Box 13-1 ISSVA Classification of Vascular Anomalies: 2014**Vascular Tumors**

- Benign
 - Infantile hemangioma
 - Congenital hemangioma
 - Tufted angioma
 - Pyogenic granuloma
 - Others
- Locally aggressive/borderline vascular tumors
 - Kaposiform hemangioendothelioma (KHE)
 - Dabska tumor
 - Kaposi sarcoma
 - Others
- Malignant vascular tumors
 - Angiosarcoma
 - Epithelioid hemangioendothelioma
 - Others

Vascular Malformations

- Simple
 - Capillary malformation (CM)
 - Venous malformation (VM)
 - Lymphatic malformation (LM)
 - Arteriovenous malformation (AVM)
 - Arteriovenous fistula (AVF)
- Combined
 - CM + VM/LM/AVM
 - CM + LM + VM/AVM
 - CM + VM + AVM
 - CM + LM + VM + AVM
- Of major named vessels
 - Affecting origin, course, number, length, diameter, or valves of larger veins/arteries/lymphatics. Includes persistence of embryonic vessels.
- Associated with other anomalies
 - Klippel-Trenaunay syndrome (KTS) (CM + VM ± LM + limb overgrowth)
 - Parkes Weber syndrome (CM + AVF + limb overgrowth)
 - Servelle-Martorell syndrome (limb VM + bone undergrowth)
 - Sturge-Weber syndrome (facial + leptomeningeal CM + ocular anomalies ± bone and/or soft tissue overgrowth)
 - Maffucci syndrome (VM ± spindle cell hemangioma + enchondroma)
 - CLOVES syndrome (congenital, lipomatous, overgrowth, vascular malformations, epidermal nevi, spinal/skeletal anomalies and/or scoliosis [LM + VM + CM ± AVM + lipomatous overgrowth])
 - Proteus syndrome (CM, VM ± LM + asymmetric somatic overgrowth)
 - Phosphatase and tensin homolog (PTEN) hamartoma-tumor syndrome, previously called Cowden syndrome or Bannayan-Riley-Ruvalcaba syndrome (AVM + VM + macrocephaly, lipomatous overgrowth)

ISSVA, International Society for the Study of Vascular Anomalies.

Box 13-2 ISSVA Classification of Simple Vascular Malformations: 2014**Capillary Malformations**

- Cutaneous and/or mucosal CM (port-wine stain)
- CM with bone/soft tissue overgrowth
- CM-AVM
- Others

Lymphatic Malformations

- Common (cystic) LM—macrocytic, microcystic, mixed cystic LM
- Generalized lymphatic anomaly (GLA)
- LM in Gorham-Stout disease
- Channel-type LM
- Primary lymphedema
- Others

Venous Malformations

- Common VMs
- Familial VM cutaneomucosal (VMCM)
- Blue rubber bleb nevus syndrome
- Glomuvenous malformation (GVM)
- Cerebral cavernous malformation (CCM, different types)
- Others

Arteriovenous Malformations and Arteriovenous Fistulas

- Sporadic
- In hereditary hemorrhagic telangiectasia (HHT)
- In CM-AVM

CM, Capillary malformation; CM-AVM, capillary malformation-arteriovenous malformation; ISSVA, International Society for the Study of Vascular Anomalies; LM, lymphatic malformation; VM, venous malformation

Establishing an accurate diagnosis and using the correct terminology are vital for appropriate patient assessment and management and avoiding inappropriate treatments. Unfortunately, confusion remains throughout the medical community regarding vascular anomalies and incorrect descriptions of these diseases, and misdiagnoses are not uncommon. Most postnatal diagnoses can be made from medical history and physical examination, supplemented with diagnostic imaging (and occasionally other investigations) when diagnoses are unclear or lesions are deeply located.

Infantile hemangiomas are the most common type of vascular tumor, initially presenting a few weeks after birth. Fetal hypoxic stress is considered a likely trigger for the development of these tumors; however, placental emboli have previously been implicated, because they have pathologic features in common. *Congenital hemangiomas*, which arise in utero and are maximal in size at birth in a full-term infant, are morphologically and biologically different than infantile hemangiomas, and their cause is not well understood. *Vascular malformations* are different than infantile hemangiomas, because they are present at birth (although not always evident), show commensurate growth with the patient, and persist throughout life. Vascular malformations are caused by errors

in vascular development during embryogenesis. The entire spectrum of vascular channels (capillary, venous, lymphatic, and arterial) can be affected, resulting in *low-flow malformations* (capillary, venous, or lymphatic malformations) or *high-flow malformations* (arteriovenous malformations [AVMs] and arteriovenous fistulas [AVFs]). In high-flow malformations, shunting of blood occurs between arteries and veins without an intervening capillary bed. Causal genetic anomalies are now recognized in several vascular malformations. For example, somatic mutations of the *Tie2* gene are seen in more than 50% of the sporadic “common type” of venous malformations. Although most vascular tumors are not associated with malformations, the so-called “segmental” type of infantile hemangioma may be associated with developmental vascular and other anomalies, such as in PHACES syndrome.³

Many patients will benefit from a multidisciplinary team approach. The interventional radiologist plays an important role on the multidisciplinary team by providing treatment options for some of these patients, particularly those with selected vascular malformations. Most treatments are performed electively; however, some may be required in emergent situations, such as in infants who have life-threatening cardiac failure resulting from large intrahepatic vascular shunts from hepatic hemangiomas and AVMs.

Interventional radiologic techniques provide “keyhole” percutaneous or endovascular approaches with imaging guidance. Procedures are carried out using a general anesthetic with full sterile technique in an angiography suite. The three main techniques for treating low-flow vascular malformations are sclerotherapy, venous embolization, and endovenous laser ablation. Sclerotherapy involves the injection of a sclerosant liquid/foam using a needle directly into the vascular channels within the malformation, whereas venous embolization involves the injection of sclerosants or occlusive devices through a catheter. Most patients with low-flow malformations treated by sclerotherapy are discharged from the hospital within 4 to 6 hours after the procedure. High-flow malformations can be treated by arterial or venous catheter embolization or by direct injection into the nidus of an AVM. High-flow malformation treatments usually require a 24-hour hospital stay before discharge. The interventional radiologist works as part of a multidisciplinary team. Sometimes radiologic treatments must be combined with surgical resection and debulking procedures, as well as combined with various medical therapies to achieve optimal results.

As a physician, the interventional radiologist sees the patient before and after the procedure and provides early and late follow-up for all patients. For elective cases, the radiologist should assess the patient and review all investigations, including recent imaging before the procedure. Arrangements are made for any necessary periprocedural, intraoperative, and postoperative care. For example, if significant swelling is likely after treatment in lesions located in the oral cavity (including the tongue) or airway, then planned admission to the intensive care unit may be needed for airway support and management. If multiple treatments are required over a long period for large anomalies affecting the oral cavity and airway (particularly in infancy), gastrostomy and tracheostomy may be required for feeding and airway support, respectively. Postoperative requirements vary and can be dependent on patient age, patient well-being (including psychosocial), and disease type and location. For example, large pelvic or urogenital malformations may require a Foley catheter for several days until postoperative swelling resolves. Steroids, antibiotics, and NSAIDs may be required as single intravenous doses for selected sclerotherapy and embolization procedures. Dexamethasone (0.1 mg/kg, maximum 8 mg) is given if the lesion is in a confined space, such as the orbit or airway and in locations where there is a risk of compartment syndrome. Cefazolin (30 mg/kg, maximum 1 g) should be given when treating oral, orbital, or perineal/perianal lesions. Ketorolac (0.5 mg/kg, maximum 15 mg in those less than 16 years of age, repeated every 6 hours) is an effective postoperative analgesic.

ROLE OF DIAGNOSTIC IMAGING

Not all patients with vascular anomalies require imaging to confirm the diagnosis. Most (more than 80%) can be diagnosed based on clinical history and physical examination; for example, most babies with infantile hemangiomas do not need imaging.⁴

Imaging is performed to aid in diagnosis and when full evaluation of the anomaly is required to assess the full extent of the disease. Ultrasonography and MRI are the most useful modalities because of their high tissue specificity.⁵ For most patients, magnetic resonance angiography (MRA) sequences are not required for diagnosis. In patients with high-flow lesions, MRA is useful for mapping the affected vascular anatomy and seeing if the lesion is embedded in fat. Ultrasonography has limitations when lesions are extensive or deeply located.

There is much “pattern recognition” on diagnostic imaging, as there is with the clinical assessment of patients, when dealing with vascular anomalies. The diagnostic radiologist must be familiar with these conditions. The radiologist should always request accurate and adequate information when the patient is referred for imaging. Knowing the patient’s age when the abnormality was first noticed and any change in lesion size as the patient has grown is vital information to help distinguish tumors from malformations. To fully evaluate the anomaly, imaging must include the entire extent of the disease. If imaging is performed to assess a hemangioma, it is useful to know the type of hemangioma (infantile/congenital), and if it is an infantile hemangioma, the phase of the life cycle of the hemangioma at the time of imaging.

Vascular tumors on MRI and ultrasonography have a parenchymal tumor showing hypervascularity and sometimes vascular shunting (Figs. 13-1 through 13-3). Depending on the tumor type and its natural history, the vascularity and shunts may vary; the degree of contrast enhancement on MRI varies and fibrofatty replacement of some tumors can occur. Tumors that are proliferative show significant hypervascularity on ultrasonography and significant contrast enhancement on MRI.

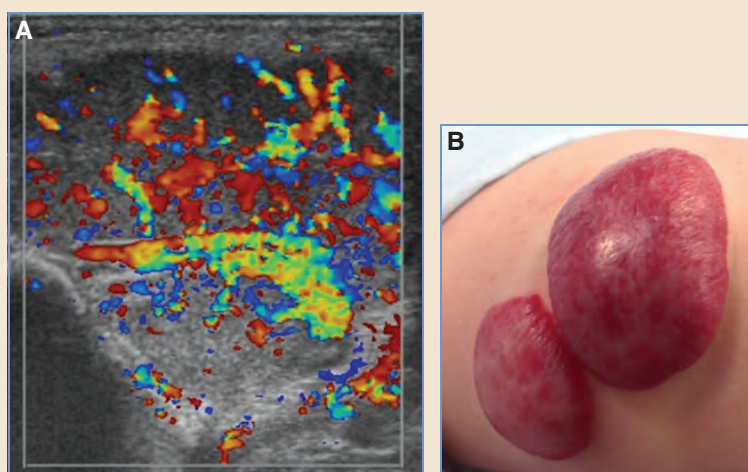


Fig. 13-1 Typical ultrasonography of infantile hemangioma. **A**, Proliferative phase of parotid infantile hemangioma. The solid echogenic tumor parenchyma is hypervascular with a high vessel density. The solid parenchymal component of the infantile hemangioma distinguishes it from an AVM. **B**, Early involutory phase of two cutaneous infantile hemangiomas.

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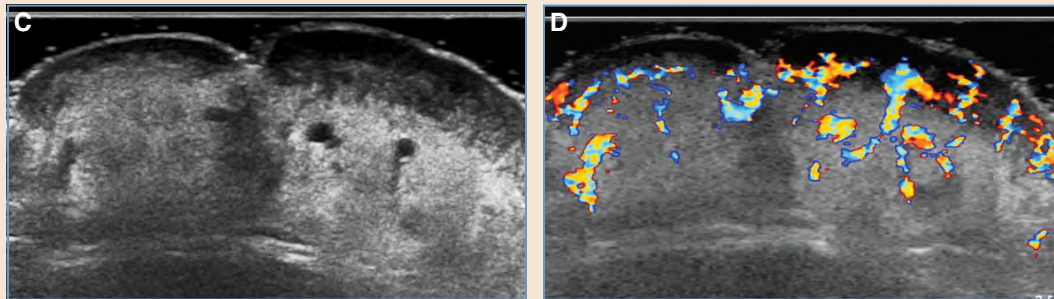


Fig. 13-1, cont'd C, Increased reflectivity to the base of the lesion in the subcutaneous tissues resulting from fibrofatty changes. D, Reduced hypervascularity and vessel density during involution on color Doppler.

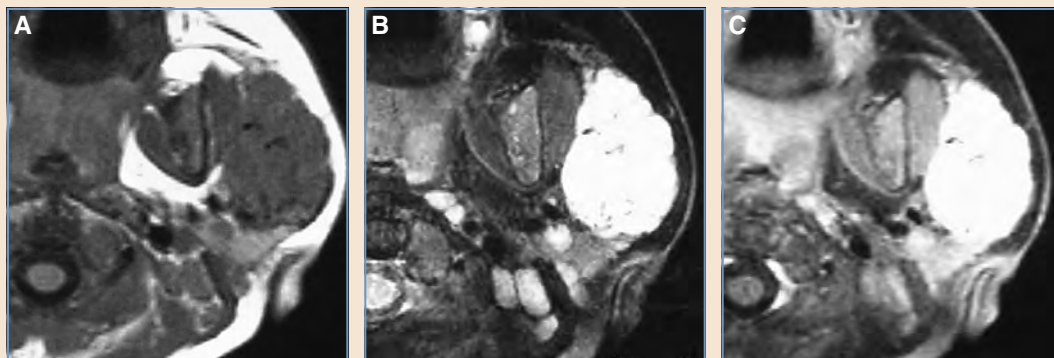


Fig. 13-2 Typical MRI features of the proliferative phase of infantile hemangioma. A, Parotid hemangioma shows that tumor parenchyma is isointense on T1-weighted sequence. B, Hyperintense on T2-weighted sequence. C, Avid uniform contrast enhancement.

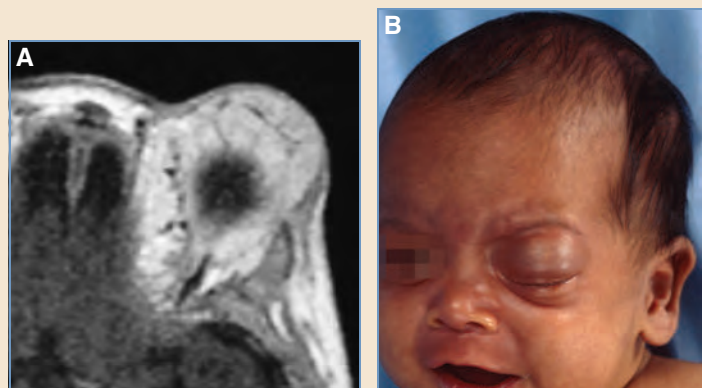


Fig. 13-3 Orbital proliferative infantile hemangioma. A, MRI shows typical flow voids in a proliferative tumor in a baby. Flow voids are seen as small channels of signal loss. B, Orbital hemangioma.

Vascular malformations on MRI and ultrasonography show no parenchymal tumor. Venous malformations have dysplastic venous spaces, thrombi, and phleboliths (Figs. 13-4 and 13-5). Lymphatic malformations have variable-sized cysts; some contain hemorrhagic fluid or thrombi adjacent to the cyst walls (Figs. 13-6 and 13-7). The microcystic type of lymphatic malformations have cysts so small that the cyst cavities cannot be identified on imaging and appear as areas of “solid” tissue, which is echogenic on ultrasonography. Venous malformations and lymphatic malformations can have fluid-fluid levels. Contrast enhancement during MRI is useful to distinguish venous malformations from lymphatic malformations. AVMs show lesional hypervascularity and vascular shunting on ultrasonography and MRI. Perilesional soft tissue edema can be seen on MRI.

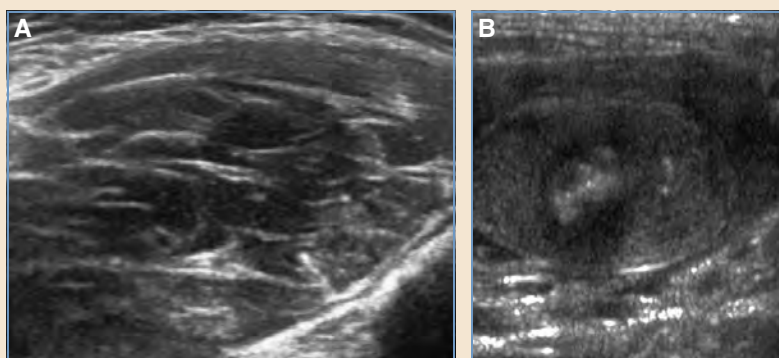
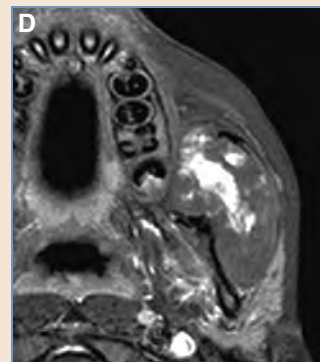
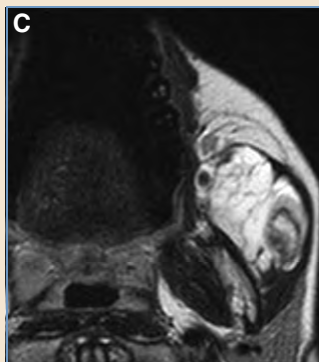


Fig. 13-4 Typical ultrasonography features of soft tissue venous malformation. **A**, Anechoic venous channels with the lesion, which has a spongeliike structure. **B**, Typical phlebolith (calcification in thrombus) present in venous malformation channel.



Fig. 13-5 Typical MRI features of venous malformation. **A**, Left facial intermittent swelling with normal overlying skin. **B**, The intramuscular venous malformation is isointense on T1 sequence. Intralesional hyperintense focal area before contrast is caused by intralesional thrombus. **C**, Hyperintense on T2 sequence. **D**, Nonuniform lesion enhancement with IV contrast is seen.



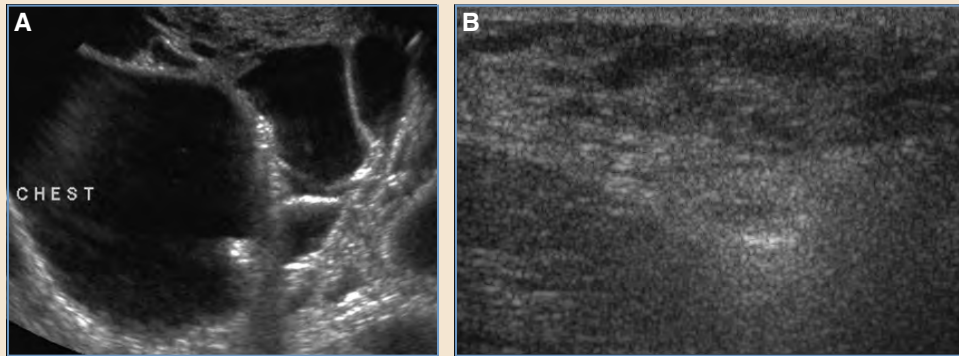


Fig. 13-6 Typical ultrasonographic features of cystic lymphatic malformation. **A**, Macrocystic lymphatic malformation. **B**, Microcystic lymphatic malformation. Cystic cavities of varying size are easily seen on macrocystic disease and are suitable for injection sclerotherapy. Pure microcystic lymphatic malformation appears as solid echogenic tissue with no ultrasonography visible cysts and is unsuitable for injection sclerotherapy.

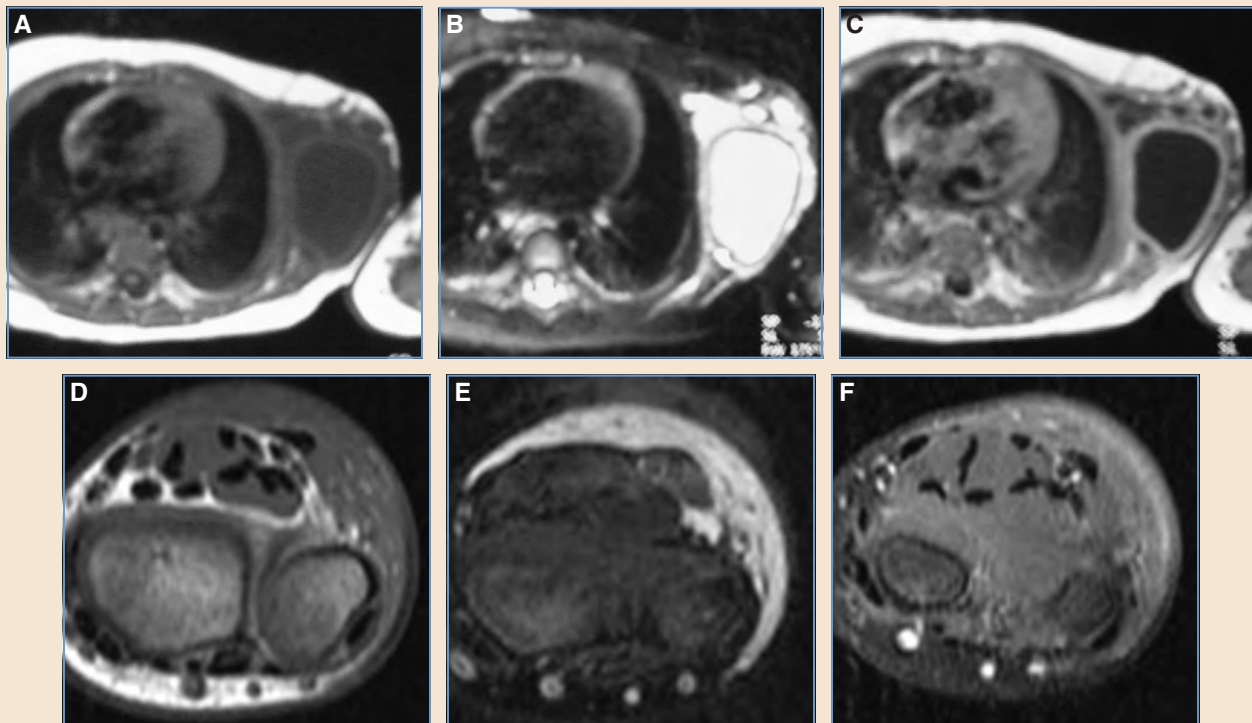


Fig. 13-7 Typical MRI features of cystic lymphatic malformation. **A-C**, Macrocystic chest wall lymphatic malformation. **D-F**, Microcystic lymphatic malformation of the forearm in the same patient shown in Fig. 13-6, *B* and Fig. 13-20, *E*. **B**, Both macrocystic and microcystic lymphatic malformations have similar T1 (**A** and **D**) and T2 (**B** and **E**) signal intensity (isointense on T1, hyperintense on T2). IV contrast can differentiate macrocystic from microcystic lymphatic malformation. **C**, Cyst rim enhancement occurs in macrocystic lymphatic malformation. **F**, Microcystic lymphatic malformation sometimes shows a faint diffuse enhancement throughout the lesion.

Localized increased fatty areas can be seen around some soft tissue venous malformations and AVMs. Enlargement of inflow arteries and outflow veins with AV shunting can be seen in both vascular tumors and high-flow vascular malformations.

Bedside ultrasonography is extremely useful for assessing vascular anomalies in a clinic setting and provides a rapid diagnosis when there is a clinical dilemma. For example, if a young infant has a slightly blue-colored soft tissue swelling, bedside ultrasonography can yield an immediate answer if it is caused by a subcutaneous infantile hemangioma, a venous malformation, or by bleeding into a small macrocystic lymphatic malformation. This is operator dependent and relies on the operator's familiarity with the ultrasonographic appearances of these disorders. This bedside ultrasound assessment is no substitute for more formal imaging in a radiology department. The interventional radiologist is well suited to carry out such bedside ultrasonography when needed in the clinic setting.

Plain radiographs and CT scans are not required for most diagnoses, because they have low-contrast resolution and tissue specificity. However, they may be required to assess abnormalities of skeletal growth associated with vascular malformations, to assess extremity overgrowth such as in Klippel-Trenaunay syndrome (KTS) and to evaluate vascular malformations involving the skeleton (Fig. 13-8). Conventional venography and catheter angiography are not required in most patients. Venography is occasionally performed in venous malformations with significant

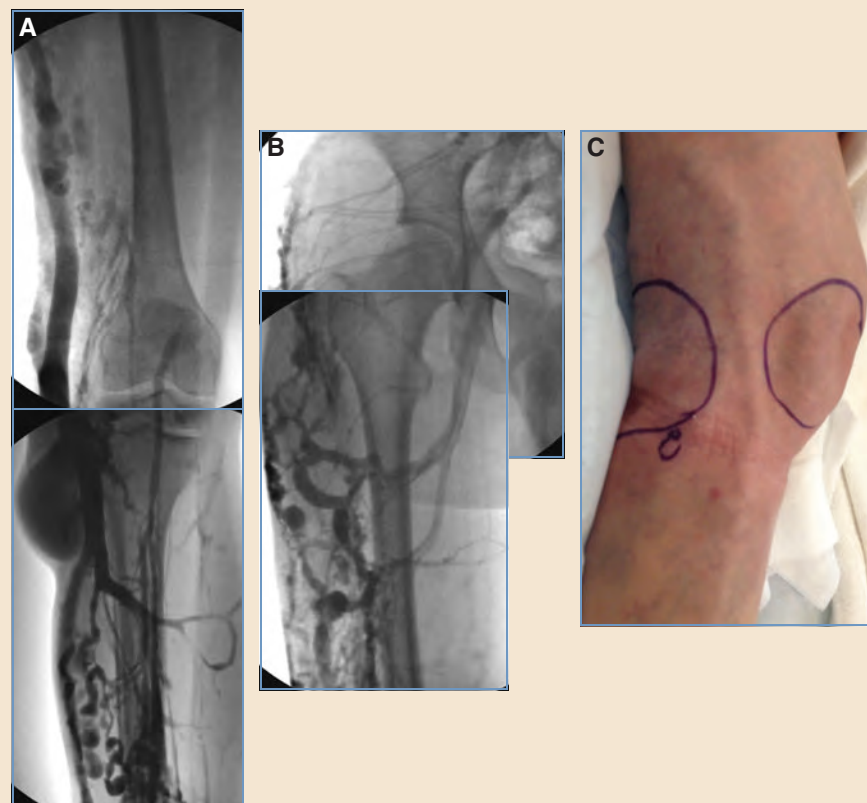
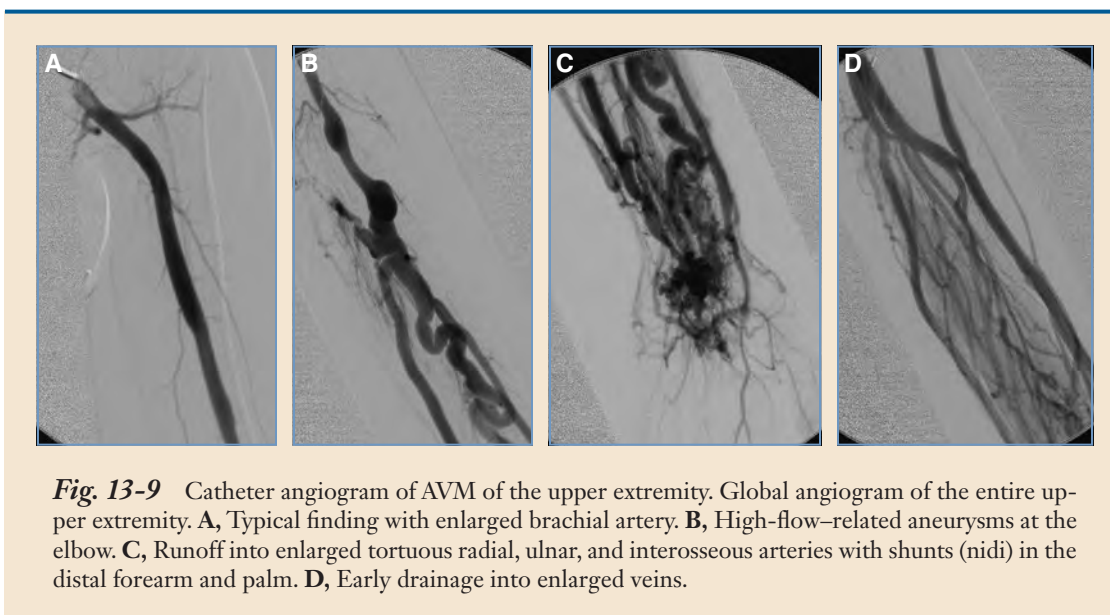


Fig. 13-8 A-C, Lateral marginal embryonic vein in KTS. **A** and **B**, Venogram of the vein performed by direct injection into the vein below the knee showing a single large channel, communicating with a superficial venous aneurysm below the knee. **B**, The embryonic vein drains through posterior channels in the upper thigh to the femoral vein and transgluteal channels to the iliac vein. This embryonic vein is frequently made up of multiple intercommunicating channels.



phlebectasia to assess potential closure methods and in some patients to delineate communications between the venous malformation and normal functioning veins. This can be performed at the time of sclerotherapy. In KTS, venography to assess anomalous veins and the deep venous system before endovascular therapy is required because magnetic resonance venography (MRV) is often inadequate. Catheter arteriography is performed during embolization in selected high-flow vascular anomalies (Fig. 13-9). Nuclear medicine has no role in imaging these disorders.

VASCULAR TUMORS

Infantile Hemangioma

Infantile hemangioma is the most common tumor of infancy, appearing a few weeks after birth and located in the skin, often in the head and neck area.⁶ Infantile hemangiomas have an incidence of 1.0% to 2.6% in white neonates, which increases significantly in preterm babies. Twenty percent of babies have multiple lesions; when internal, the lesions can be present in the liver, gastrointestinal tract, and brain. Infantile hemangiomas have a characteristic three-phase life cycle (see Fig. 13-9). The first or *proliferative phase* occurs within the first 9 to 12 months of postnatal life and is characterized by rapid tumor growth and tumor angiogenesis. During this phase tumors are hypervascular and vascular shunting can be seen. The second or slow *involuting phase* occurs between 1 and 5 to 7 years of age, with reduced angiogenesis and reduction in overall size. Reduced vascular flow and some fibrofatty replacement occurs. The third or *involved phase* occurs after 5 to 7 years, with resultant fibrofatty residue, small capillaries, and draining veins (Fig. 13-10). After involution, lesions do not recur, and approximately half of all patients will have near-normal looking skin at the site of the lesion.

When the superficial dermis is involved, infantile hemangiomas present as red lesions and become raised. Ulceration and bleeding can be seen in large superficial tumors with rapid growth. Tumors in the lower dermis and subcutaneous tissue appear bluish or have no alteration in color to the overlying skin. During involution, the color of the tumor fades.

Infantile hemangiomas can be mistaken for capillary malformations. Deep (for example, subcutaneous) infantile hemangiomas can be mistaken for venous malformations or lymphatic malformations because of the bluish discoloration through the skin. Infantile hemangiomas showing

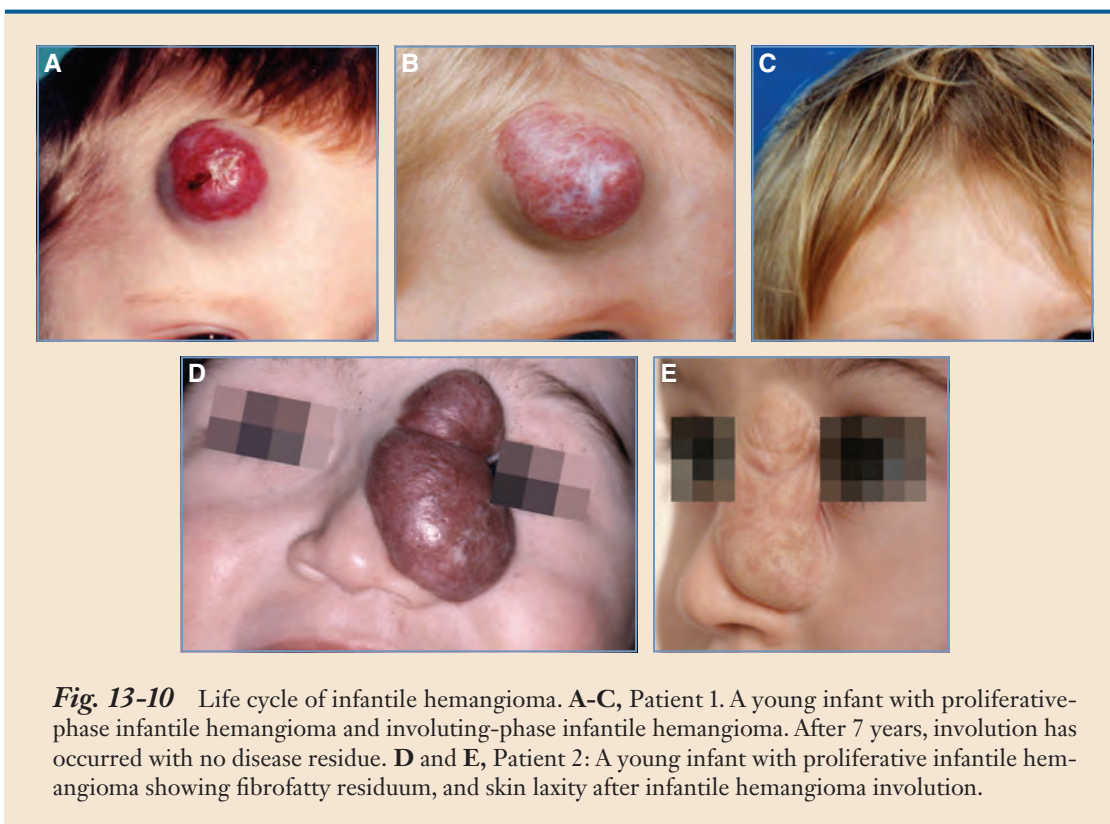


Fig. 13-10 Life cycle of infantile hemangioma. **A-C**, Patient 1. A young infant with proliferative-phase infantile hemangioma and involuting-phase infantile hemangioma. After 7 years, involution has occurred with no disease residue. **D** and **E**, Patient 2: A young infant with proliferative infantile hemangioma showing fibrofatty residuum, and skin laxity after infantile hemangioma involution.

high flow can be mistaken for AVMs, but the age at onset, growth pattern, and presence of tumor parenchyma in an infantile hemangioma distinguishes it from an AVM.

The location of the infantile hemangioma may be associated with specific problems during the proliferative phase when the tumor is rapidly increasing in size. Airway obstruction, visual impairment, gastrointestinal bleeding, high-output congestive heart failure from vascular shunting within large or multiple hemangiomas (particularly in the liver), and tissue necrosis can potentially occur.

Imaging of Infantile Hemangioma

On ultrasonography, infantile hemangiomas are well-defined masses during the proliferative phase with a nonspecific echogenicity, which is either hyperreflective or hyporefective (see Fig. 13-1). High vessel density, high Doppler frequency shifts, and low resistive indexes are characteristic of proliferative infantile hemangiomas⁷ (see Fig. 13-1). During involution, the lesion becomes smaller with decreased vessel density. However, vessels usually maintain high Doppler frequency shifts. Fibrofatty changes, especially at the base of infantile hemangiomas, occur with involution (see Figs. 13-2 and 13-3 for the typical MRI appearance of proliferative infantile hemangioma).

Congenital Hemangioma

Congenital hemangiomas are rare and are distinct from infantile hemangiomas.^{8,9} They arise antenatally and are fully developed at birth. They do not usually exhibit postnatal growth unless infants are preterm, when they will continue to grow for some time. There are three recognized types of congenital hemangiomas: (1) *rapidly involuting congenital hemangioma* (RICH), (2) *non-involuting congenital hemangioma* (NICH, Figs. 13-11 through 13-14), and (3) *partially involuting*

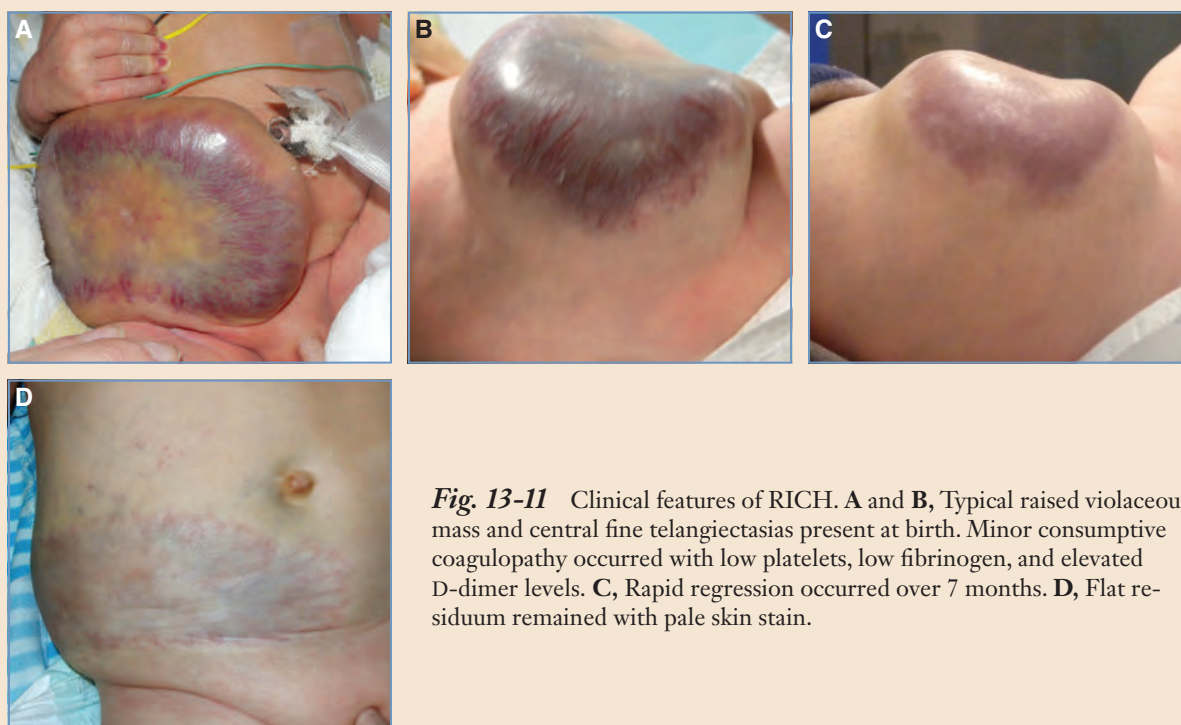


Fig. 13-11 Clinical features of RICH. **A** and **B**, Typical raised violaceous mass and central fine telangiectasias present at birth. Minor consumptive coagulopathy occurred with low platelets, low fibrinogen, and elevated D-dimer levels. **C**, Rapid regression occurred over 7 months. **D**, Flat residuum remained with pale skin stain.

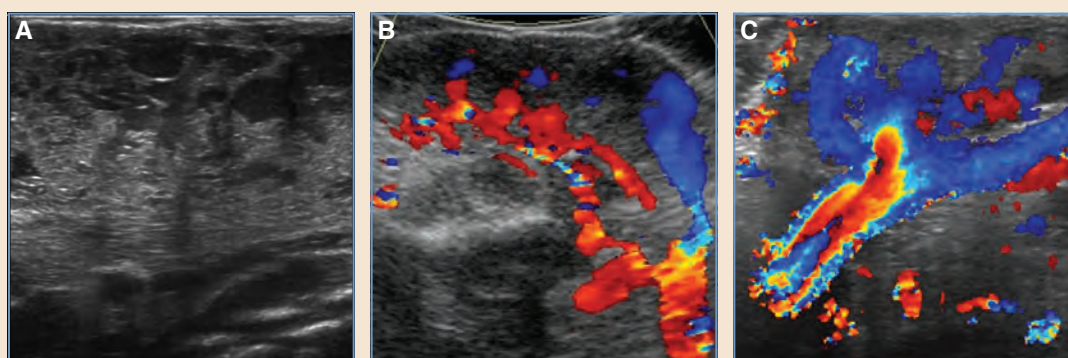
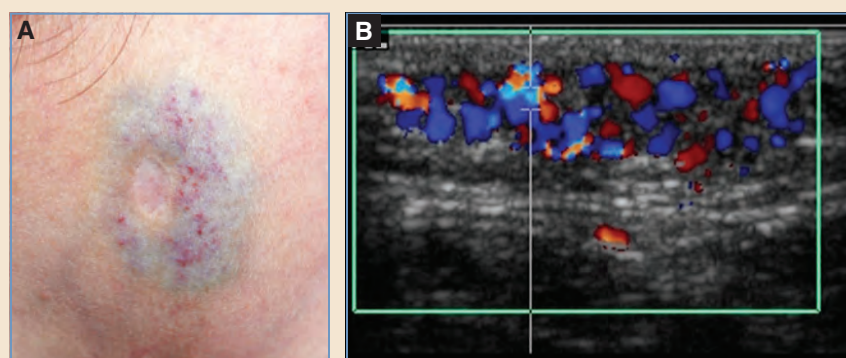
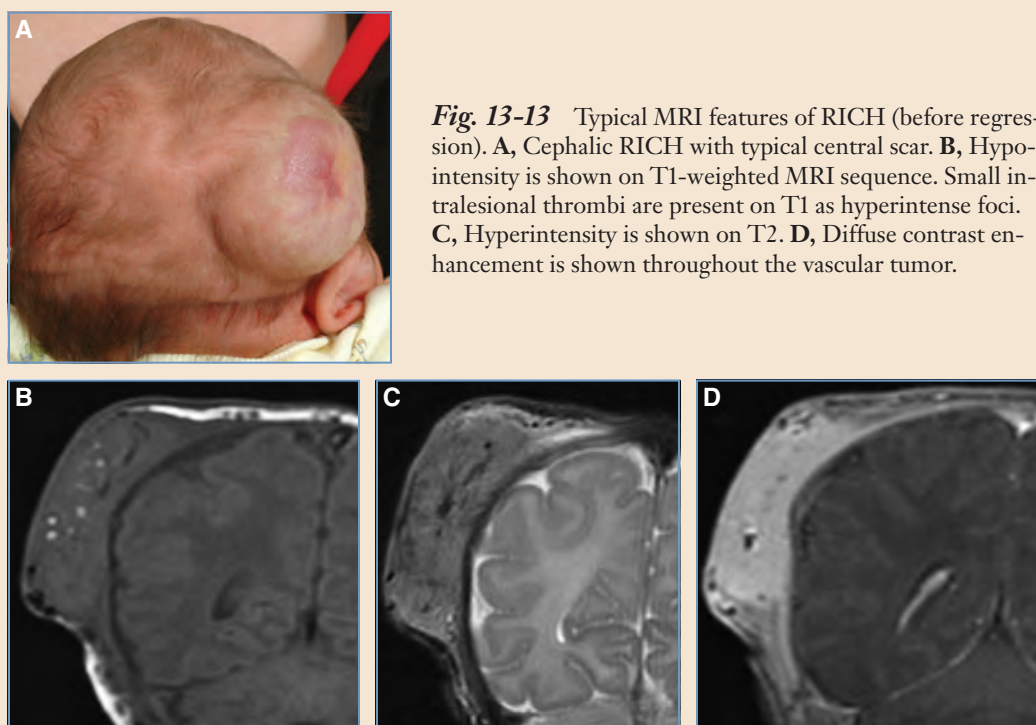


Fig. 13-12 Typical ultrasonography features of RICH (before regression). **A**, Typical vascular soft tissue tumor containing anechoic vascular spaces. **B** and **C**, Marked hypervascularity with prominent arterial inflow and venous outflow on color Doppler imaging.

congenital hemangioma (PICH). Clinically, these three types of congenital hemangiomas are quite different. RICHs are often impressive protuberant masses, with a predilection for the craniofacial region and extremities, whereas NICHs tend to be flat to slightly raised, rounded skin lesions, with telangiectasias and a pale surrounding halo. RICHs show spontaneous accelerated regression between 6 and 14 months of postnatal life, whereas NICHs persist throughout life. Before regression, two complications can be seen in the RICH, although both are uncommon: cardiac overload and transient thrombocytopenia. Significant arteriovenous shunting can occur within the tumor, leading to cardiac failure. The thrombocytopenia typically occurs in the first



week in large RICH tumors, and platelet count rapidly returns to normal levels as the RICH regresses. A PICH has overlapping features of a RICH and a NICH; however, even though it can resemble a RICH initially, it never fully regresses. Congenital hemangiomas can potentially be confused with vascular malformations. However, the clinical features and the presence of tumor parenchyma provide clues to the antenatal and postnatal diagnosis of congenital hemangiomas. These tumors, although called “hemangiomas,” are quite distinct from infantile hemangioma. Interestingly, hemangiomas may be seen with features in common with both tumor types (infantile hemangioma and congenital hemangioma) in the same lesion.

Imaging of Congenital Hemangioma

Distinctive imaging characteristics are observed in a congenital hemangioma.⁹ On ultrasonography, prominence of vascular channels and calcifications are seen in contrast to infantile hemangioma. On MRI, the tumor margins may be less defined compared with infantile hemangioma⁹ (see Figs. 13-12 through 13-14).

Infantile Hepatic Hemangioma

Visceral hemangiomas may be associated with multiple cutaneous infantile hemangiomas (more than five). However, they can occur without cutaneous hemangiomas. The liver is the most common organ affected by hemangiomas. The three types of *infantile hepatic hemangiomas* (IHHs) are focal, multifocal, and diffuse¹⁰ (Fig. 13-15). The classic presentation of IHH with the symptoms of heart failure, anemia, and hepatomegaly is rare, and certainly not all hepatic hemangiomas are life-threatening. Most infantile hepatic hemangiomas, even if multifocal, are asymptomatic.

Focal lesions are often large and present antenatally. They do not grow after birth, with regression usually seen in the first 12 to 14 months of postnatal life (similar to a RICH). Multifocal hepatic hemangiomas are similar in behavior to the typical cutaneous infantile hemangiomas with a three-phase life cycle. Macrovascular shunts, if present in the focal and multifocal IHHs, can lead to high-output cardiac failure and require emergent catheter embolization, which is sometimes performed as staged procedures.¹⁰⁻¹² In focal IHH, the onset of cardiac failure is usually at or soon after birth. In multifocal IHHs, the onset of cardiac failure may not occur for several months after birth. Embolization should be performed in conjunction with other intensive medical support and medical therapy (including steroids and beta-blockers, the latter being used with caution in cardiac failure) to promote tumor involution. In the diffuse type, most of the liver parenchyma is replaced by tumor, typically causing massive hepatomegaly, abdominal compartment syndrome, and respiratory compromise. The diffuse type can lead to severe hypothyroidism until there is tumor involution because the tumor produces type 3 iodothyronine deiodinase, which inactivates circulating thyroid hormones. Large thyroxine doses are required by these infants.

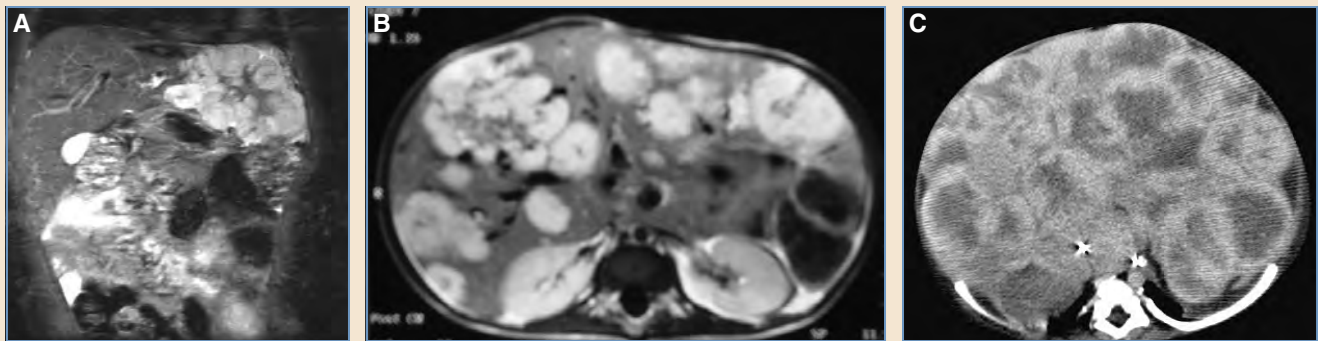


Fig. 13-15 IHH. The three types of IHH are shown: **A**, Focal. **B**, Multifocal. **C**, Diffuse. **A** and **B** are T2-weighted MRI sequences, whereas **C** is a postcontrast CT scan.

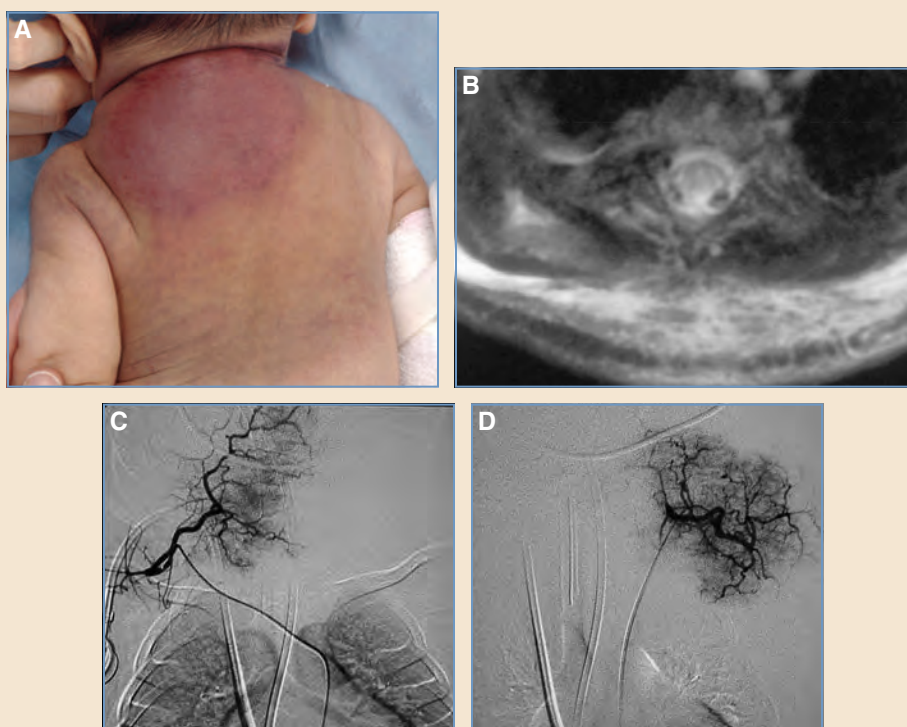


Fig. 13-16 KHE. **A**, Typical appearance of KHE with violaceous tumor. **B**, MRI (T2-weighted sequence) shows a hyperintense soft tissue tumor with typical subcutaneous stranding from dilated subdermal lymphatics. **C** and **D**, Selective microcatheter angiography shows typical hypervascular tumor with no shunting.

Kaposiform Hemangioendothelioma and Tufted Angioma

These sometimes aggressive and invasive vascular tumors are often present at birth. However, they can also present later. *Kaposiform hemangioendothelioma* (KHE) is usually more extensive than *tufted angioma*. The tumors are often located on the trunk, shoulder, thigh, or retroperitoneum.¹³ Tufted angioma appears as a red skin plaque and KHE as a violaceous skin lesion. MRI is excellent at evaluating these rare vascular tumors (Fig. 13-16). Tissue biopsy is not always required; however, a percutaneous image-guided needle biopsy can be helpful. Histologic findings reveal infiltrating sheets of slender endothelial cells with positive staining for lymphatic elements.

There is no role for interventional radiology in the treatment of tufted angioma or uncomplicated KHE.

Kasabach-Merritt Syndrome

Kasabach-Merritt syndrome refers to thrombocytopenia from platelet consumption associated with a coagulopathy and occurs only with two specific vascular tumors, KHE and tufted angioma. It does not occur in other vascular tumors or any vascular malformation.

Typically, the thrombocytopenia is severe, with a platelet count below $10 \times 10^9/L$. The coagulopathy occurs with reduced fibrinogen levels and elevated prothrombin and partial thromboplastin times. Bleeding is often evident. Treatment for most patients is medical, because tumors

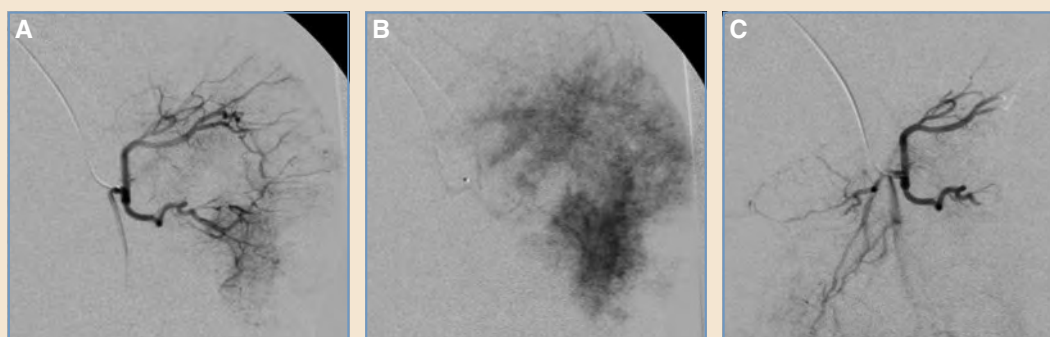


Fig. 13-17 Catheter embolization of KHE. **A** and **B**, Typical angiographic appearance of KHE with tumor staining. During arterial catheter embolization, particles (for example, polyvinyl alcohol) are used to devascularize the tumor parenchyma. **C**, Angiographically, particles are delivered to reduce the tumor stain and preserve the main arterial trunks.

may be large and extensive.^{14,15} Supportive measures in critical care may be needed. Low-dose weekly IV vincristine and oral corticosteroids are usually given. More recently, rapamycin (sirolimus) has also been used effectively. In Kasabach-Merritt syndrome, the mortality rate is between 20% and 30%, and tumors show proliferation into early childhood with eventual incomplete regression despite being otherwise asymptomatic. Platelet transfusions are usually reserved for patients with platelet counts below $10 \times 10^9/L$ when there is a risk of spontaneous intracranial hemorrhage. Routine platelet transfusion is not recommended, because this can lead to an increase in tumor size.

Transcatheter arterial embolization with polyvinyl alcohol particles (size range 250 to 350 μ) can improve the thrombocytopenia (Fig. 13-17) and may need to be repeated. Particle embolization should be considered adjunctive therapy if the thrombocytopenia is refractory to medical treatment with pharmacologic agents.

Vascular Tumor Treatment

Most infantile hemangiomas do not require specific treatment other than observation and reassurance to the family. In particular, the interventionalist has no role in the majority of infants with vascular tumors.

Previously, approximately 10% of infantile hemangiomas were treated medically, because they were “endangering” by virtue of their location or functional risks (for example, airway compromise or heart failure). Today, more infants with cutaneous infantile hemangiomas are offered medical treatment during the proliferative phase. Oral beta-blockers (propranolol) or angiogenesis inhibitors, mainly corticosteroids (prednisone), are commonly used. Topical beta-blockers are now being used in selected lesions. Intralesional corticosteroid injection may be performed for infantile hemangiomas that cause local deformity or ulceration. Ultrasonographic guidance can help to target the injection into the solid tumor component. Systemic low-dose chemotherapy with vincristine (through a central venous line or a peripherally inserted central catheter line) may be considered additional therapy in challenging cases.

In selected patients, other options can be offered, such as surgery with excision and debulking. Surgical resection is sometimes performed for infantile hemangiomas during and after the involuting phase and also for NICHs. An Nd:YAG laser with epidermal ice cooling has been recommended as an option to treat the deeper subcutaneous component of infantile hemangiomas.

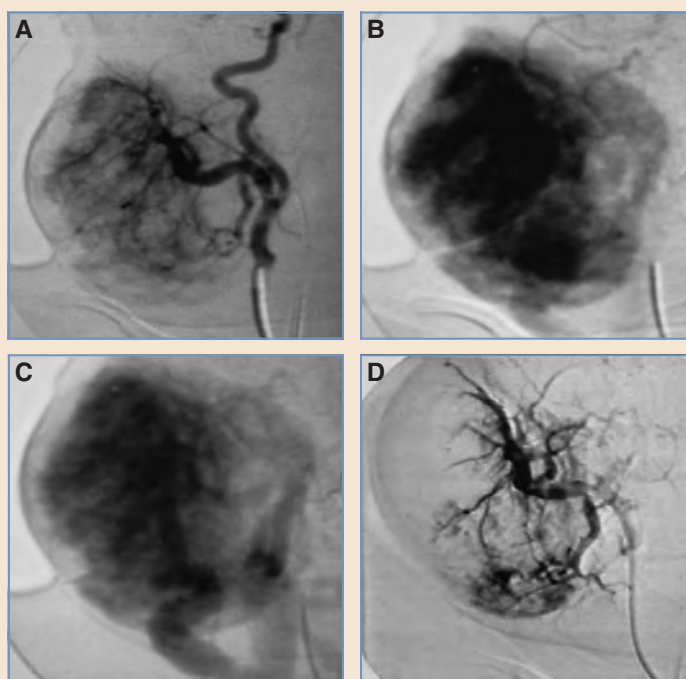


Fig. 13-18 Catheter angiography and embolization in proliferative phase infantile hemangioma. An infant with a large parotid infantile hemangioma. Typical angiographic findings. **A**, Enlarged feeding facial artery. **B**, Marked tumor parenchymal staining. **C**, Enlarged draining external jugular vein typical of proliferative infantile hemangioma. **D**, Particle embolization results in significant devascularization. NOTE: Most infants with infantile hemangioma do not require catheter angiography and embolization.

It is important to wait for complete regression of RICHs and completed involution in infantile hemangiomas before plastic surgery is considered to deal with cosmetic issues.

Catheter angiography and embolization is rarely needed for infants with vascular tumors. However, it can be extremely useful and sometimes lifesaving when endovascular closure of macroshunts or devascularization of the tumor bed is required (see Fig. 13-17; Fig. 13-18).

Catheter embolization is a useful technique in a few selected infants in the following clinical situations:

1. To reduce the tumor vascular shunting when infants are in cardiac failure
2. To devascularize the tumor if there is refractory tumor bleeding
3. To devascularize the tumor in Kasabach-Merritt syndrome when it is not responsive to conventional medical management
4. To devascularize the tumor before surgical excision, if there are concerns about intraoperative blood loss

Angiography and catheter embolization in infancy require much expertise. Arterial catheterization can be challenging in infancy, because of small access vessel sizes. In a full-term infant, the common femoral artery has a diameter of 2 mm, and the diameter of the brachial artery is approximately 1.5 mm. It is essential to use small angiographic catheters (for example, 3 Fr or 4 Fr), safe volumes of radiologic contrast, and systemic heparinization during catheterization to minimize catheterization times and to maintain patient well-being, including body temperature and fluid balance. Familiarity with vascular anatomy and variable shunts is essential. Because staged

embolization may be required, it is important to avoid routine diagnostic catheter angiography in infants unless it is combined with embolization to preserve vital arterial access. Embolic agents can be safely delivered through a microcatheter, which is advanced to the target through an outer 4 Fr guiding catheter. Embolic agents used to treat vascular tumors include N-butyl-2 cyanoacrylate (NBCA) glue (Histoacryl), polyvinyl alcohol microparticles, microcoils, and occasionally small vascular plugs. Ethanol and sodium tetradecyl sulphate (STS) should not be used as embolic agents for vascular tumors, because they can cause extensive tissue necrosis.

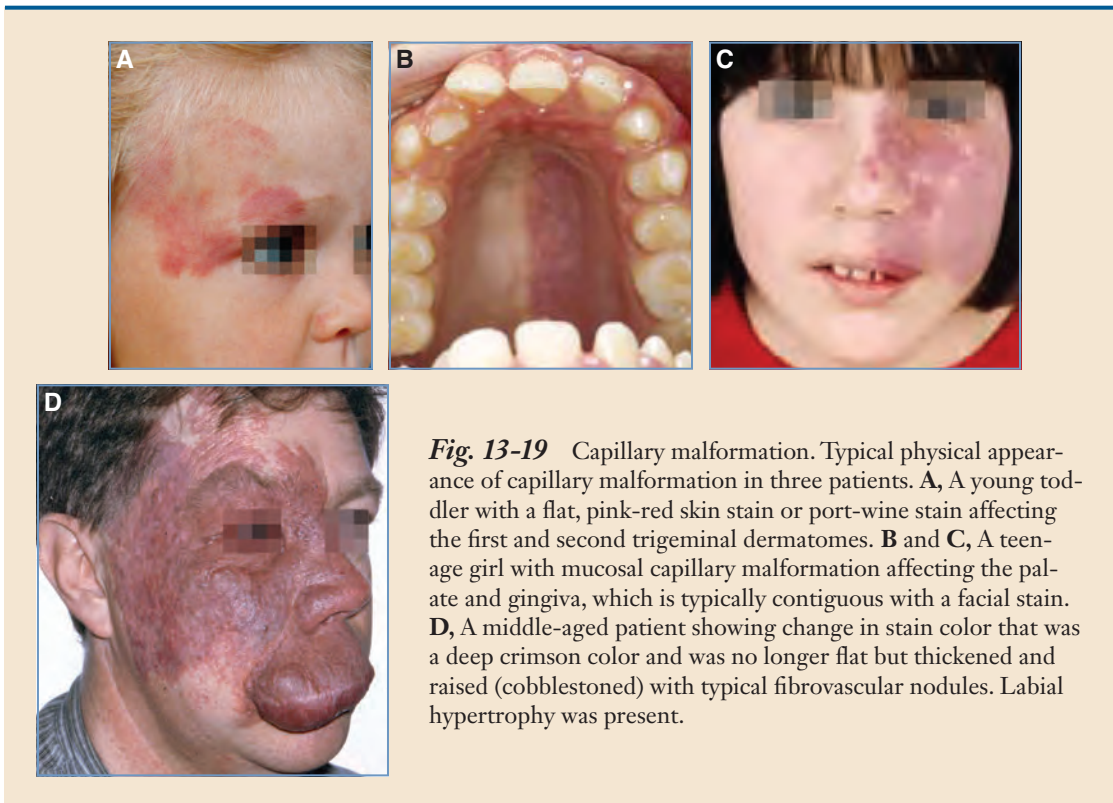
VASCULAR MALFORMATIONS

Vascular malformations are quite rare, with an estimated incidence of approximately 0.3%. They are typically sporadic (with no family history); however, they can sometimes occur as incompletely penetrant, inherited traits. The genetic basis of several malformations has been identified, and to date, genetic advances have predominantly been seen in the rare, familial forms.

Capillary Malformation

A *capillary malformation* is the most common vascular malformation and at birth is a flat, pink-red, low-flow cutaneous lesion. Capillary malformations are commonly called *port-wine stains* and frequently are located as focal skin stains in the head and neck area (Fig. 13-19). They can also be mucosal. Capillary malformations can be localized or extensive. They do not fade and are permanent (see Fig. 13-19).

Syndromic capillary malformations occur more rarely, such as in *Sturge-Weber syndrome* (SWS), in which the capillary malformation is located on the face and affects the first and sec-



ond trigeminal dermatomes (see Fig. 13-19). Sturge-Weber syndrome is associated with ipsilateral leptomeningeal vascular malformations and cerebral atrophy. In the complex combined low-flow vascular malformation, KTS, the capillary malformation commonly affects a lower extremity. The diagnosis of a capillary malformation is clinical, and no imaging is required to diagnose it. Treatment of capillary malformations is for cosmetic appearance; flash-lamp pulsed dye laser therapy is the treatment of choice.¹⁶ There is no role for interventional radiology or surgery in the treatment of capillary malformations.

Common (Cystic) Lymphatic Malformation

Common (cystic) lymphatic malformations are made up of cysts containing lymphatic fluid that are low flow and are classified as macrocystic, microcystic, or mixed cystic lymphatic malformations.¹⁷ Although the definition of macrocysts and microcysts remains undecided, a good practical definition is based on macrocysts having cavities visible on imaging, whereas microcysts appear as “solid” tissue with cysts seen only histologically. However, some consider macrocysts as those cysts greater than 1 cm in diameter and microcysts as less than 1 cm. Outdated terminology, such as *cystic hygromas* and *lymphangiomas*, should no longer be used, because they imply “tumor.”

Common (cystic) lymphatic malformations often present as localized, sometimes infiltrative masses, although some can present with fluid leakage, such as chylous fluid in body cavities or weeping from skin vesicles (Fig. 13-20). The cystic type of lymphatic malformations are usually noted at birth; however, they can be seen at any age, including prenatally on imaging. Although skin and soft tissues (subcutaneous and fascial planes in muscle) are most commonly affected, lymphatic malformations can involve bone and more rarely viscera such as the gastrointestinal tract and lungs. The axillary, thoracic, cervicofacial, mediastinal, retroperitoneal, buttock, and anogenital regions are commonly affected. Soft tissue and skeletal hypertrophy can occur with lymphatic malformations. Macrocystic lymphatic malformations appear as soft, partially compressible masses. Macrocystic lymphatic malformations, unlike common venous malformations, show no increase in size or dependency and no refilling after compression is released. Lymphedema can occur if there is diffuse infiltration of subcutaneous tissues. Interestingly, infection and

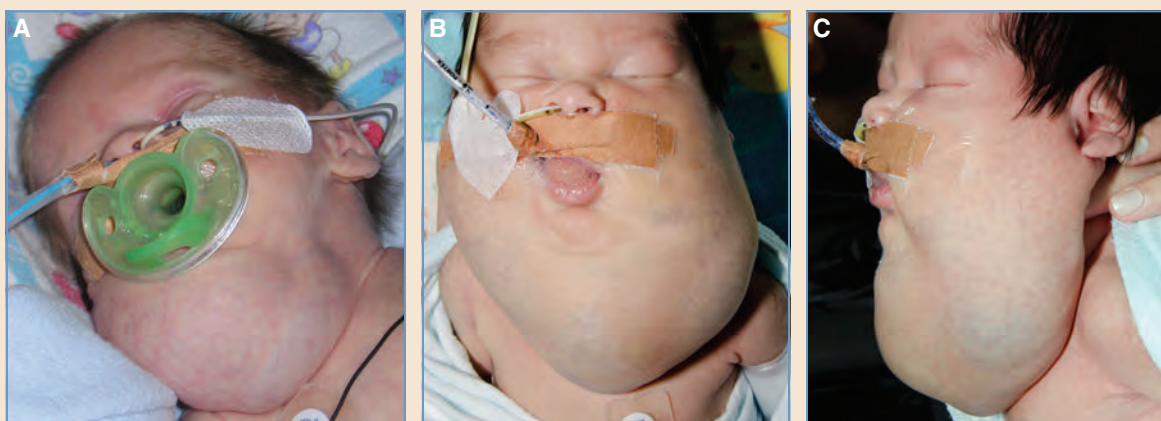


Fig. 13-20 Clinical features of cystic lymphatic malformations. Typical findings in five patients. **A**, Newborn with large, cervical, partially compressible macrocystic lymphatic malformation. **B** and **C**, Newborn with a large mixed cystic lymphatic malformation.

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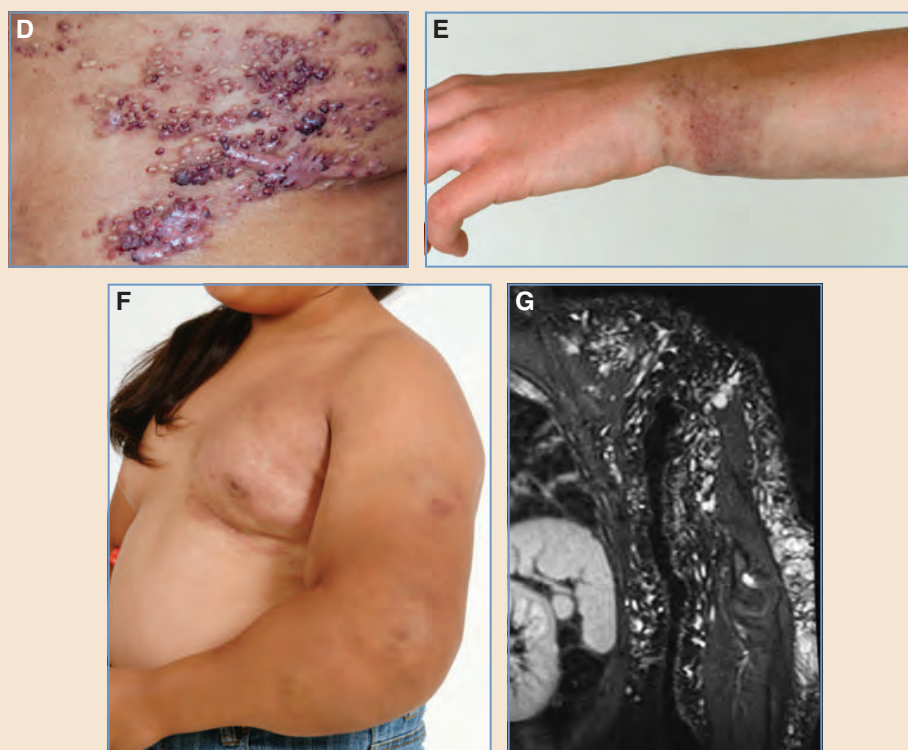


Fig. 13-20, cont'd **D**, Microcystic lymphatic malformation of the skin (sometimes referred to as *lymphangioma circumscriptum*) with recurrent bleeding from skin lesions. **E**, Firm, noncompressible expansion of the wrist with bruising (from spontaneous intralesional hemorrhage) in a microcystic lymphatic malformation (the same patient is shown in ultrasonography scan in Fig. 13-6, *B*, and in MRI scan in Fig. 13-7, *D-F*). **F** and **G**, Extensive subcutaneous microcystic lymphatic malformation in the upper extremity, shoulder, and chest wall.

intralesional bleeding can occur in all types of cystic lymphatic malformations; this sometimes leads to alarming expansion of the lesion with pain and a red/blue skin discoloration.

The problems associated with cystic lymphatic malformations can frequently be related to their location, such as proptosis from periorbital lymphatic malformations, airway obstruction in cervicofacial lymphatic malformations, oozing of fluid and bleeding from cutaneous and lingual lymphatic malformation vesicles, and protein-losing enteropathy in gastrointestinal tract disease. Lymphatic malformations of the extremities can be associated with overgrowth and limb-length discrepancy. In *Gorham-Stout disease* (soft tissue and skeletal lymphatic malformations leading to progressive osteolysis), pathologic fractures and vertebral instability can occur.

Imaging of Cystic Lymphatic Malformation

Cystic lymphatic malformations are best imaged by ultrasonography and MRI⁴ (see Figs. 13-6 and 13-7), particularly before starting treatment. Imaging determines the macrocystic and microcystic disease and helps in treatment planning. MRI better documents the full extent of deeper, larger, and more complex lymphatic malformations; however, ultrasonography can answer many questions before treatment for many patients. Macrocystic structures are easily visible on ultrasonography and MRI, especially when cysts are larger than 1 cm in diameter. Microcystic lymphatic malformations appear as predominantly solid lesions with no identifiable cysts on imag-

ing. Cystic lymphatic malformations on MRI appear hypointense on T1 and hyperintense on T2 (because of high water content). Macrocystic lymphatic malformations show septal contrast enhancement, whereas microcystic lymphatic malformations show either no contrast enhancement or ill-defined enhancement of the entire lesion. Fluid-fluid levels in cysts are caused by layering of protein or blood. Enlarged or anomalous venous channels can sometimes be seen in close proximity to cystic lymphatic malformations. Contrast lymphangiography is not required for patients with cystic-type lymphatic malformations; however, contrast lymphangiography is useful in selected lymphatic malformations in which there are abnormal lymphatic channels and leaks of the thoracic duct and in patients with chylous effusions.

Treatment of Cystic Lymphatic Malformation

The indications for treatment of cystic lymphatic malformations include troublesome symptoms with recurrent swelling, pain, infections, functional impairment, leakage of fluid and bleeding from the lymphatic malformation, and cosmesis. Spontaneous intralesional bleeding causes sudden enlargement of cystic lymphatic malformations and is treated conservatively. Analgesics should be given for pain. Lymphatic malformation enlargement occurring with systemic viral or bacterial infections usually resolves without specific treatment. Bacterial infections occurring in the lymphatic malformation resulting in a cellulitis-like presentation with reddening, enlargement, hardening, and pain requires appropriate antibiotics (often intravenous), analgesics, and hospital admission. Some patients with frequent recurrent infections of their lymphatic malformation should be considered for long-term prophylactic antibiotics. Chronic pain in those patients with extensive disease requires expert management.

Other treatment options include injection sclerotherapy, surgical interventions, laser techniques, conservative measures, and combinations thereof.^{18,19} Recently, medical therapy with rapamycin and sildenafil has been suggested for selected patients.^{20,21} Rapamycin is now used in patients with complicated vascular anomalies when there is a lymphatic component and particularly when there are no other/limited treatment options.²¹ Macrocystic lesions can be treated through sclerotherapy or resection with excellent results in nearly 90% of patients.^{19,22} The advantages of sclerotherapy over surgical resection for macrocystic lymphatic malformations includes the avoidance of surgical scars, reduced risk of neurovascular injury, and when the disease is present in challenging anatomic locations, uneventful recoveries in most patients (Figs. 13-21 and 13-22).

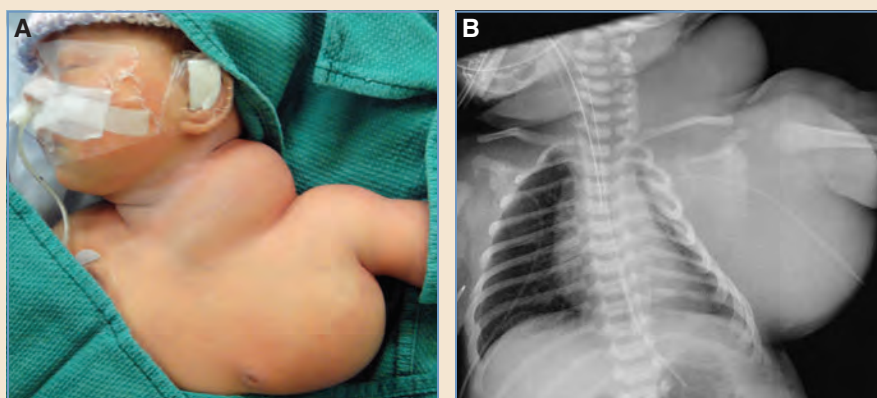


Fig. 13-21 Injection sclerotherapy and drainage procedure for large cervicothoracic macrocystic lymphatic malformations. **A** and **B**, Large lymphatic malformation distorting the thoracic cavity.

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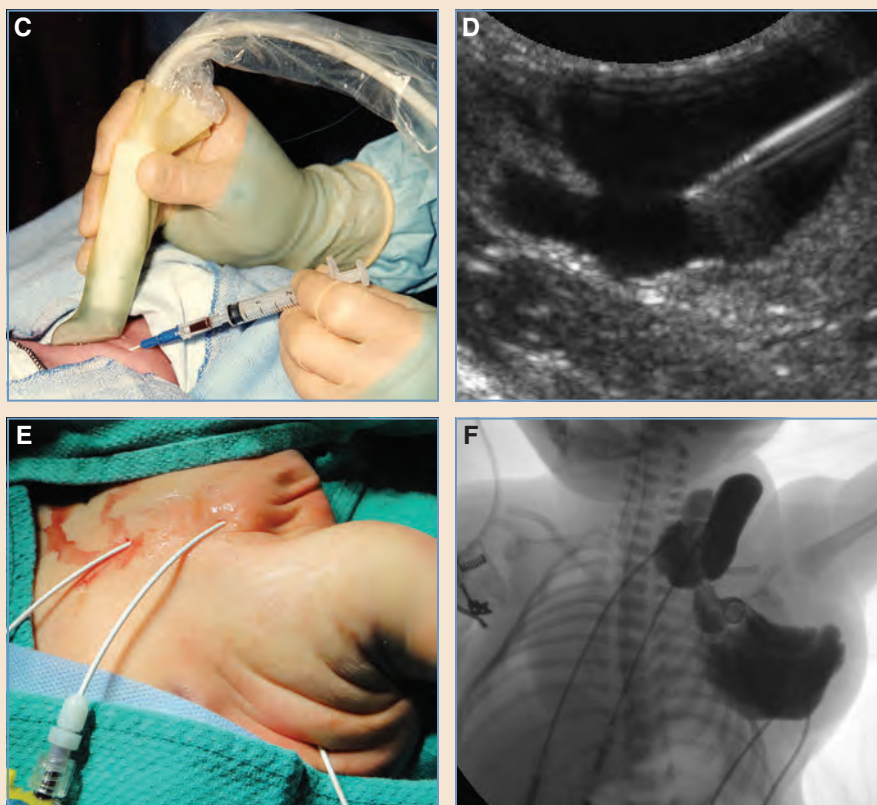


Fig. 13-21, cont'd C and D, Ultrasonographic guidance used to puncture cysts. E, Larger cysts drained and cysts aspirated before sclerosant injection. F, Contrast injected through drains to outline cysts before sclerosant injection. We no longer inject contrast for such routine cases.

For well-localized macrocystic lymphatic malformations, sclerotherapy can be curative (see Fig. 13-22), and at least 90% of patients with macrocystic lymphatic malformations will significantly improve. Diffuse and predominantly microcystic lymphatic malformations are difficult to eradicate, although sclerotherapy can offer some reduction in lesion size, depending on the number of cysts that can be injected.¹⁹

When only a few cystic cavities are seen on ultrasonography in a microcystic lymphatic malformation that is predominantly “solid” on ultrasonography, we would not routinely offer injection sclerotherapy. Surgical resection of complex and microcystic lymphatic malformation can be beneficial and usually needs to be well planned and staged. The postsurgical recurrence rate, however, for microcystic lymphatic malformations is approximately 40% and is the result of regrowth and/or reexpansion of residual disease. Multiple scars from many surgeries are not uncommon, which themselves can lead to problems. Microcystic lymphatic malformations that were deep to the skin before surgery can grow externally along scars after surgery. It has been suggested that sclerotherapy of the resection cavity may help to reduce the recurrence of disease. Injection sclerotherapy can also help temporarily to control weeping or bleeding from cutaneous vesicles.

It has been difficult to compare treatment outcomes from surgery and injection sclerotherapy. Surgical results to date have been based on cosmetic appearances, without postsurgical imaging to assess deep residual disease.

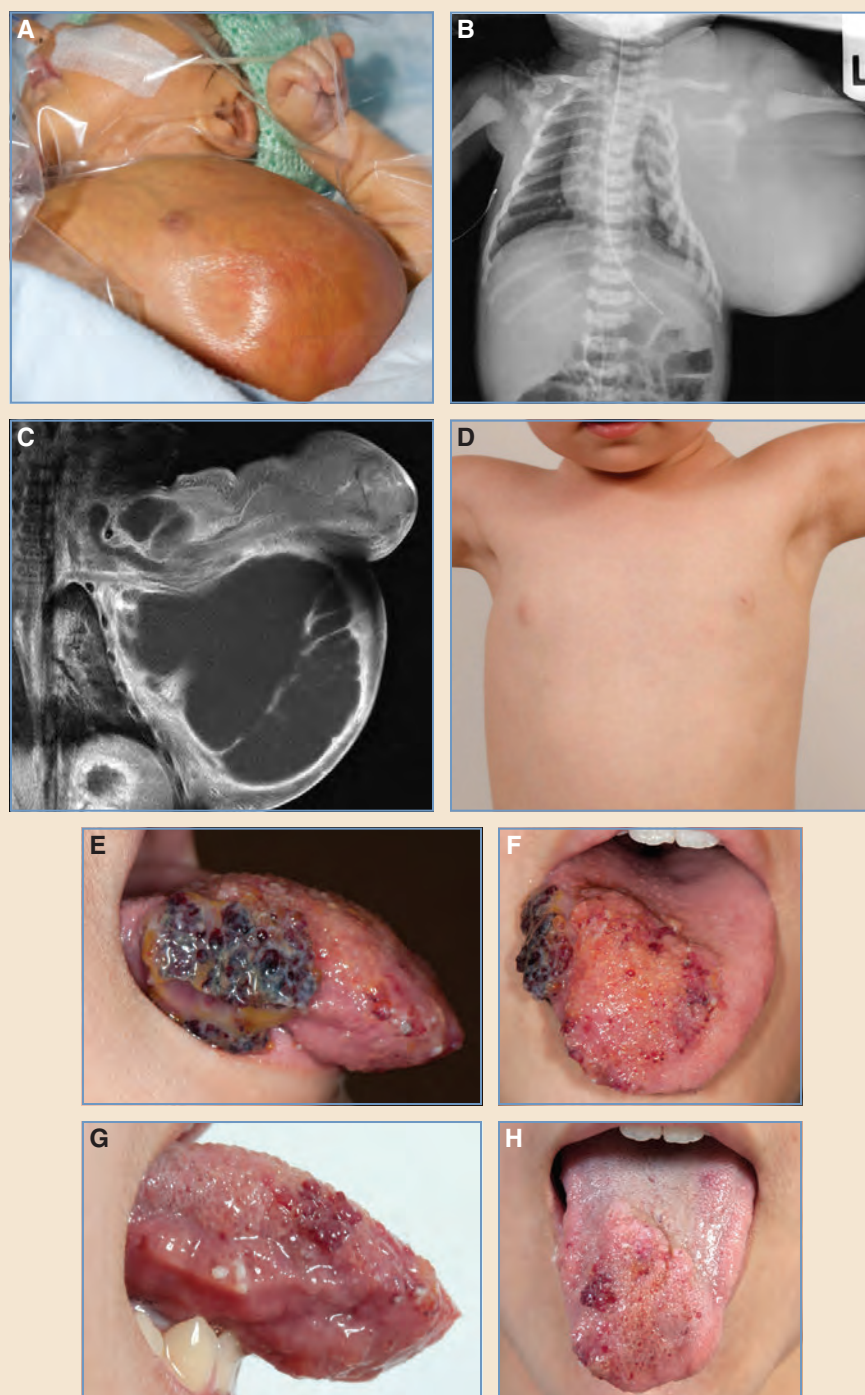


Fig. 13-22 Results of lymphatic malformation sclerotherapy for macrocystic lymphatic malformation and microcystic lymphatic malformation. **A-D**, Excellent results for macrocystic disease can be expected with administration of doxycycline. This infant with a large thoracic macrocystic lymphatic malformation had complete resolution after sclerotherapy using small drainage catheters, with no disease residue clinically on imaging. **E-H**, Lingual microcystic disease can be improved with bleomycin. After direct bleomycin injection into the tongue of this patient, there was no recurrence of bleeding, and many tongue surface vesicles regressed.

Cystic Lymphatic Malformation Sclerotherapy

Most injection sclerotherapy procedures are performed with ultrasonographic guidance (see Fig. 13-21). When sclerosing macrocysts, the cysts are aspirated and then sclerosant is injected. In our practice, for macrocysts greater than 5 cm diameter, we place small drains (5 Fr) into these larger cysts, aspirate fluid, and then inject sclerosant (see Fig. 13-21). Drains are opened after 6 hours, and repeat sclerosant injection is performed through the drain the following day before drain removal. Fluoroscopic guidance is rarely needed unless drains are placed. CT guidance may be required when drains are placed in challenging locations, such as the mediastinum and other intrathoracic locations, and for intraosseous disease. When sclerosing microcysts, it may be impossible to direct the needles into very small cysts, because often there are no cyst cavities visible. Therefore the sclerosant is injected throughout the entire lesion.

The dose of sclerosant injected into the lesion depends on the aspirate volume when treating macrocysts or the overall size of the lesion when treating microcystic disease. There are several useful sclerosants for treating cystic lymphatic malformations (Box 13-3).

Bleomycin has been shown to be effective in many patients with lymphatic malformations.²³⁻²⁸ It is associated with significantly less swelling after injection compared with other sclerosants; therefore bleomycin has a significant advantage when administered to treat lesions in the orbit, tongue, and airway (see Fig. 13-22). Dosage has been a concern with bleomycin; however, for sclerotherapy, low intralesional doses are used compared with the significantly higher systemic dosage used for chemotherapy. In our practice we restrict the use of bleomycin for treating macrocystic and microcystic lymphatic malformations of the tongue, orbit/eyelid, and airway.

After sclerotherapy, some swelling of the treated area is expected. Significant pain is unexpected and managed appropriately. Complications are unusual and unexpected after lymphatic malformation sclerotherapy. Infrequent self-limiting minor bleeding can be seen in the treated cyst or cysts during the procedure or shortly thereafter. Rarely local skin blistering is seen with doxycycline and occurs with overfilling of superficial cysts.

Most lymphatic malformation sclerotherapy is performed as an elective day-case procedure with a 4-hour postprocedure stay. Overnight or intensive care admission may be required and arranged as necessary before treatment. At discharge, the patient should be systemically well

Box 13-3 Sclerosants for Treating Cystic Lymphatic Malformations

For Macrocystic Lymphatic Malformations

- Doxycycline solution is used in a 10 mg/ml concentration in sterile water, with a maximum patient dose between 300 mg for babies and 1200 mg for children 12 years of age and older. Half the volume of cyst aspirate is replaced with doxycycline solution.
- 3% STS liquid is used to irrigate larger macrocysts (greater than 5 cm) before doxycycline injection. The maximum dose of STS is 0.5 ml/kg body weight.
- Ethanol or dehydrated alcohol 100% is our second-line sclerosant after doxycycline for macrocysts. The maximum patient dose is 1 ml/kg.

For Microcystic Lymphatic Malformations

- Bleomycin is used for those patients with solid disease on imaging in selected locations (tongue, eyelid, orbit, or airway) in a concentration of 1 mg/ml. A suggested maximum patient dose is 0.5 mg/kg for neonates and 1 mg/kg or 15 mg (15 units) for older patients.
- Doxycycline (as above), only if small cystic cavities visible on ultrasonography can be injected.

with no concerns regarding the treatment site other than expected swelling and occasional skin bruising. Intralesional sclerotherapy results in scarring and cyst collapse and may need to be repeated at 6-week intervals. Arrangements should be made for appropriate posttreatment clinical and imaging follow-up.

Venous Malformation

Venous malformations are the most frequent type of vascular malformation referred to vascular anomaly clinics and the “common type” are the most frequent venous malformations treated by pediatric interventional radiologists. They are low-flow lesions and consist of sinusoidal venous spaces with thin walls, abnormal smooth muscle cells, and variable communications with adjacent veins. In a *glomovenous malformation* (GFM, a variant of venous malformation), the vascular channels are lined by glomus cells.²⁹ Venous malformations can arise anywhere in the body and may involve all tissue planes (Fig. 13-23).



Fig. 13-23 Clinical features of common venous malformation. Four patients with typical physical findings. **A**, Large periscapular venous malformation with blue compressible swelling and a network of dilated veins. **B**, Venous malformation of the hand with blue compressible swelling of the palm and the second through fifth fingers. The hand swellings were reduced in size through compression/arm elevation and increased in size on release of compression/placement of the limb in a dependent position. **C**, Diffuse venous skin stain of the lower extremity signaled diffuse intramuscular venous malformation throughout the leg. **D**, Reduced girth of the right leg from a diffuse intramuscular venous malformation in the thigh and calf is associated with osteoarticular disease of the knee joint. There is no skin stain in the affected leg in this patient.

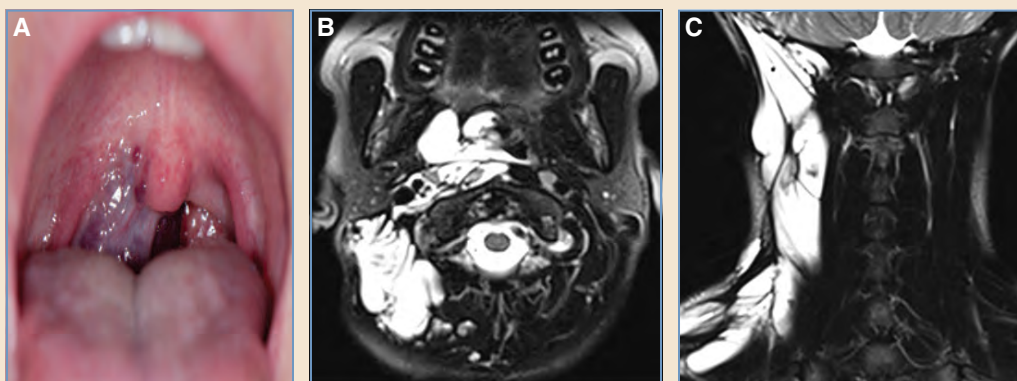


Fig. 13-24 Orocervicopharyngeal venous malformation. **A**, A teenage girl with hemoptysis from a right-sided oropharyngeal venous malformation. Clinical examination showed obvious right-sided blue venous malformation. The full extent of the disease was not appreciated until the MRI scan was done, because she had no symptoms related to her neck and no cervical abnormality on physical examination. **B** and **C**, MRI showed an extensive soft tissue venous malformation in the oropharynx abutting the airway, with intramuscular extension throughout the right neck.

Venous malformations are often seen in the skin and soft tissues.^{4,30} Forty percent of them affect the head and neck, 40% affect the extremities, and the remainder affect the trunk. Visceral involvement can occur. They grow in proportion to the patient and show no regression. Venous malformations may be evident at birth or more commonly become apparent and sometimes symptomatic with pain and swelling later in childhood. Their appearance is variable, including varicosities, phlebectasias, discrete spongy masses, or complex channels permeating tissues and organs (Figs. 13-24 and 13-25). Most occur sporadically and are unifocal, although some are multifocal or diffuse. Diffuse venous malformations commonly affect an entire extremity with much muscle and large joint involvement, particularly the knee joint (see Fig. 13-25). On physical examination, they appear as soft, bluish, compressible lesions, showing refilling with release of compression and increasing in size with dependency or during a Valsalva maneuver. As many as 42% of patients with a venous malformation have a localized intralesional coagulopathy, which is severe in approximately 4% and seen particularly in those with diffuse intramuscular involvement, where significant elevated D-dimer and low fibrinogen levels can be detected in peripheral blood.³¹⁻³³ Intralesional thrombosis occurs and is sometimes associated with acute pain. Muscle wasting is evident in those with diffuse disease. Involvement of bones and joints may lead to pathologic fractures, recurrent hemarthroses and arthritis with osteoarticular changes similar to hemophilia (see Fig. 13-25). In patients with a diffuse type of venous malformation, pulmonary hypertension has been recently reported. However, it is unclear if this is caused by recurrent silent pulmonary emboli.³⁴

Venous malformations, which are often multiple, can occur throughout the gastrointestinal tract. Venous malformations of the pelvis and perineum are associated with venous malformations in the descending colon and rectum. Gastrointestinal bleeding, typically chronic, can cause anemia. In blue rubber bleb nevus syndrome, multifocal venous malformations are seen in the skin (typically on the palms of the hands and soles of the feet) and soft tissues, in addition to the gastrointestinal tract. The diagnosis of gastrointestinal involvement is best made by endoscopy, including wireless capsule endoscopy. If bleeding is problematic, surgical and endoscopic treatment options should be considered for gastrointestinal venous malformations.

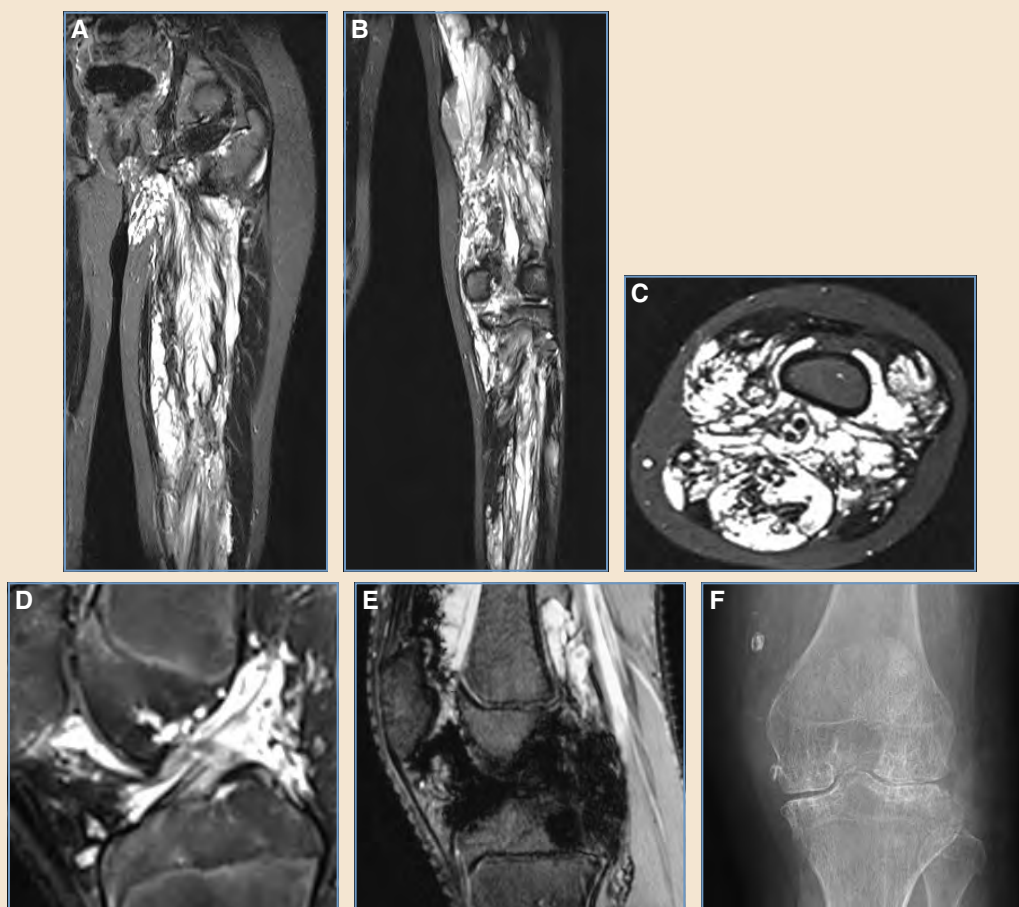


Fig. 13-25 Diffuse lower extremity venous malformation. **A-C**, T2-weighted MRI sequences show diffuse intramuscular involvement throughout the lower extremity with intraarticular venous malformation. **D**, Cruciate ligaments are enveloped by the articular venous malformation. **E**, Gradient echo MRI sequence shows hemarthrosis. Hemosiderin appears as artifact with black areas of signal loss on gradient echo sequences. **F**, Severe arthropathy can result from recurrent hemarthroses.

Imaging of Common Venous Malformation

Common venous malformations are best imaged by ultrasonography and MRI⁵ (see Figs. 13-4 and 13-5). Ultrasonography can assess superficial and focal lesions. Ultrasonography typically shows a localized lesion with anechoic channels that are compressible and show refilling with release of pressure. Variable solid elements in the lesion and sometimes hyperreflective phleboliths (calcified thrombi) with acoustic shadowing can be seen on ultrasonography. MRI is highly tissue specific, with T1 hypointensity, T2 hyperintensity, and nonuniform lesional enhancement after contrast. The T2 hyperintensity is caused by slow-flowing blood in the venous malformation. The T1 intralesional high signal is caused by thrombi. Phleboliths within venous malformations are seen as signal voids on T2 imaging. Phleboliths distinguish venous malformations from tumors and all other vascular malformations. The pattern of contrast enhancement distinguishes venous malformations from cystic lymphatic malformations. MRI shows the full extent of the venous malformation, because there may be significant disease not evident by clinical examination

(see Figs. 13-23 and 13-24). In patients with diffuse extremity disease, MRI is needed to assess intraarticular involvement, and to evaluate the full extent of the disease (see Fig. 13-24).

Invasive venography is not required before treatment. Direct percutaneous puncture venography is performed at the time of injection sclerotherapy. If conventional venography of functional veins is required, it can be performed at the time of sclerotherapy.

Treatments of Common Venous Malformation

The indications for treatment include troublesome symptoms with pain, swelling, functional impairment, bleeding (hemarthroses, gastrointestinal, and genitourinary), and cosmesis. Treatment options include injection sclerotherapy, laser techniques (usually endovenous), surgical interventions (excision and partitioning), endoscopic treatments, conservative measures, and combinations thereof.³³ Combining sclerotherapy with surgical techniques can be advantageous in selected patients.

Direct puncture intralesional sclerotherapy is the mainstay of treatment for many common venous malformations.^{4,35,36} However, if the venous malformation is small, focal, and superficial, surgical excision may be preferable. When postinjection surgical excision is planned, glue injection into venous malformations is an alternative to sclerosant injection.³⁷ Sclerosants cause an intense early inflammatory response that can potentially be challenging for the surgeon.

For diffuse venous malformations of the extremities, conservative management with graded compression stockings can achieve significant symptom improvement if they are correctly fitted, deliver adequate compression, are worn daily, and are replaced regularly (three to four times per year). It is now recommended that early surgery be performed in patients with intraarticular venous malformation, even if asymptomatic, to remove the articular venous malformation and prevent joint destruction.³⁸ In patients with troublesome pain from intralesional thrombosis (where D-dimer levels are elevated greater than three times normal), low-molecular-weight heparin can be considered and needs to be given by subcutaneous injection twice daily for several weeks, which often results in excellent pain relief. Compression garments can improve the abnormal coagulation indices.

Patients with facial venous malformations are commonly present for sclerotherapy treatment because of disfigurement, whereas those with extremity venous malformations are usually treated because of pain. Although most venous malformation treatment is not curative, many patients gain symptomatic relief. Most published series document improvement in pain after sclerotherapy.^{39,40} As many as 90% of patients expect to see some reduction of pain. However, it is well recognized that pain tolerance differs among patients. The worst outcomes for pain improvement after sclerotherapy are in patients with venous malformations involving an entire muscle or muscle compartment.

Common Venous Malformation Sclerotherapy

Preprocedural blood is drawn to establish D-dimer and fibrinogen baseline levels, and this should be performed in all patients, especially those with diffuse venous malformations. If the fibrinogen level is less than 1 g/L, low-molecular-weight heparin is required before treatment to improve the effectiveness of sclerotherapy and reduce intraprocedural bleeding. It is important to be aware that both sclerotherapy and surgical excision of common venous malformations can change a preexisting localized intralesional coagulopathy into a disseminated intravascular coagulopathy with potential life-threatening bleeding.

Most procedures are performed with ultrasonographic guidance (Fig. 13-26). If sclerotherapy is limited to small cutaneous channels, direct puncture without image guidance may be sufficient. CT guidance is rarely needed for venous malformation sclerotherapy unless challenging loca-

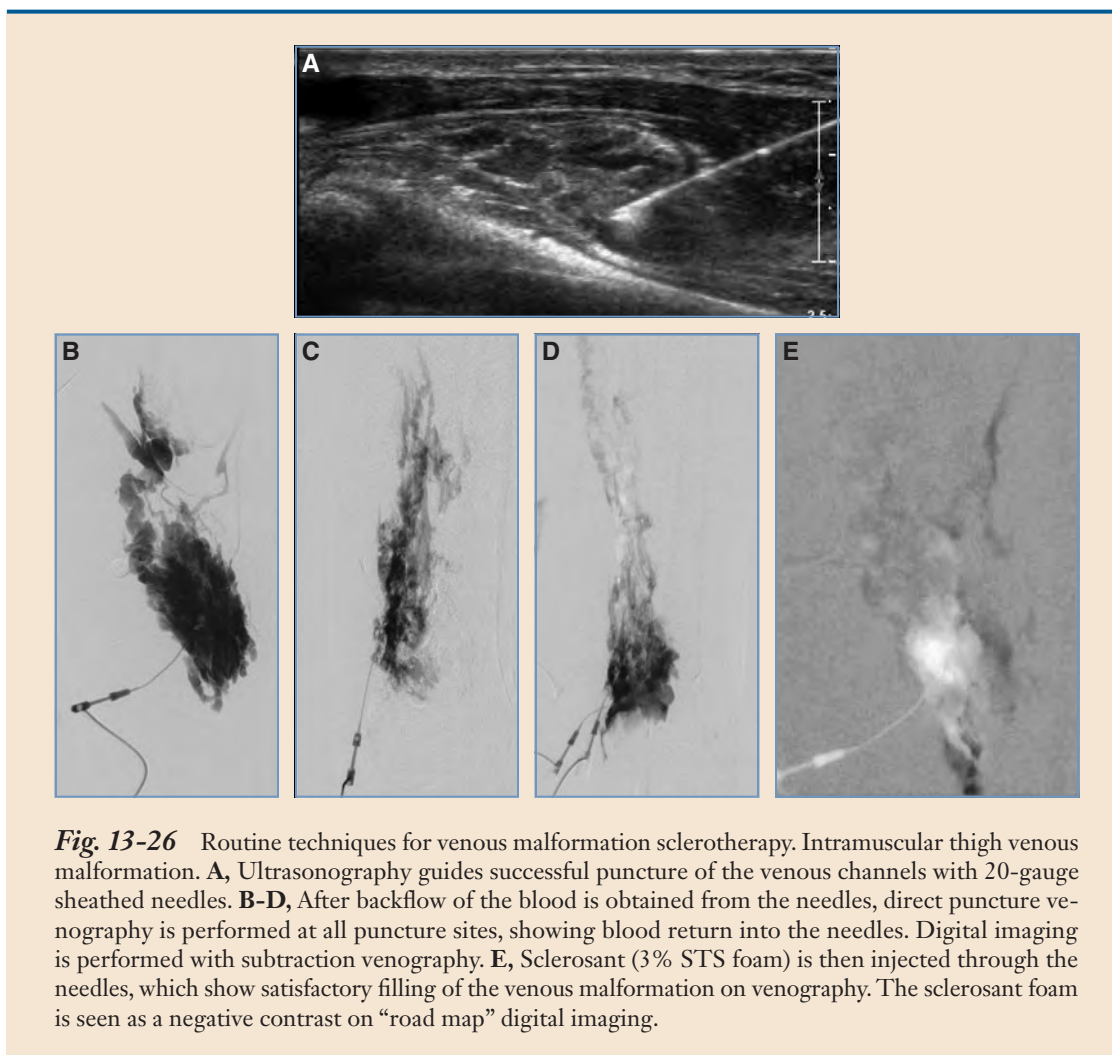


Fig. 13-26 Routine techniques for venous malformation sclerotherapy. Intramuscular thigh venous malformation. **A**, Ultrasonography guides successful puncture of the venous channels with 20-gauge sheathed needles. **B-D**, After backflow of the blood is obtained from the needles, direct puncture venography is performed at all puncture sites, showing blood return into the needles. Digital imaging is performed with subtraction venography. **E**, Sclerosant (3% STS foam) is then injected through the needles, which show satisfactory filling of the venous malformation on venography. The sclerosant foam is seen as a negative contrast on “road map” digital imaging.

tions are treated, which may occur with mediastinal, intrathoracic, or intraosseous disease. To enlarge a venous malformation of the extremity to aid in puncture, an external tourniquet is applied immediately above the venous malformation and is always removed before venography and sclerosant injection. The venous malformation is punctured, with appropriate multiple needles placed throughout the venous malformation (see Fig. 13-26; Fig. 13-27). Direct puncture venography performed at each puncture site should show satisfactory filling of the malformation, and then sclerosant is injected at suitable sites (see Fig. 13-26).

Sclerosing agents used for intralesional sclerotherapy into venous malformations include 3% STS foam, ethanol, and bleomycin. STS and ethanol cause endothelial damage, thrombosis, scarring, and a reduction in the size of the venous malformation. However, 3% STS has gained popularity, because intra-procedural fatalities and major systemic complications are associated with ethanol. The maximal doses at any one treatment should not exceed 0.5 ml 3% STS liquid or 1 ml ethanol/kg body weight. STS can be easily made into a foam by agitating an equal volume of liquid and air. Potential complications associated with the use of STS and ethanol are covered in Box 13-4.

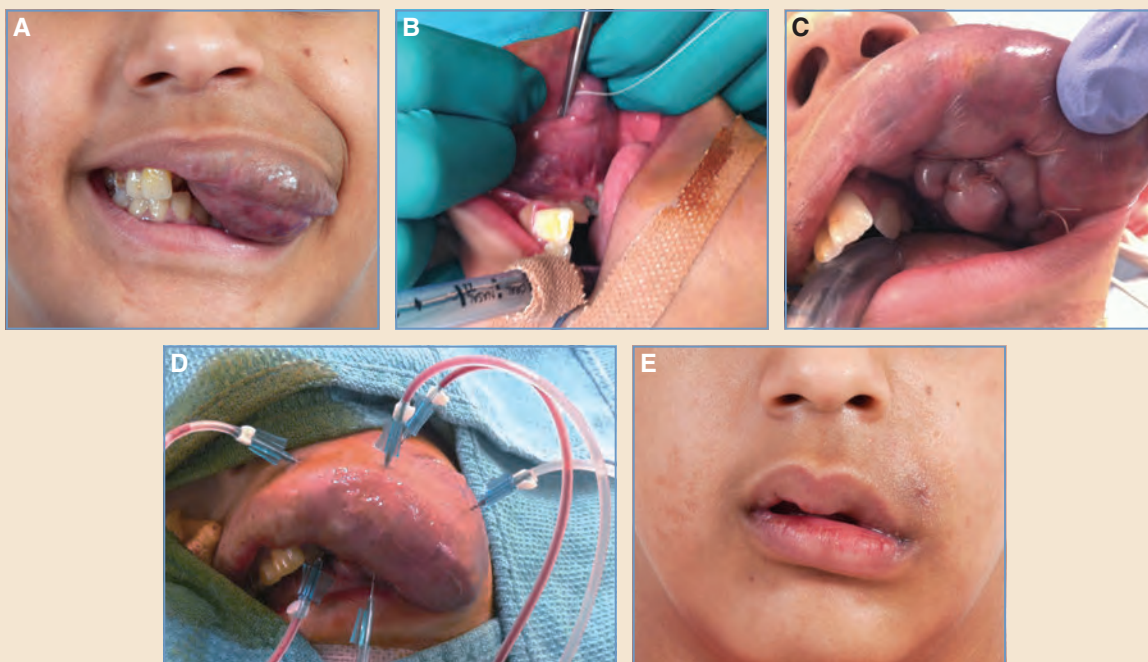


Fig. 13-27 A, Combined surgical partitioning and injection sclerotherapy of a soft tissue venous malformation. Prominent upper lip venous malformation. B and C, The venous malformation was partitioned by suturing the lesion with Vicryl sutures. D, Multiple 23-gauge butterfly needles were then placed throughout the partitioned venous malformation. Good backflow of blood was obtained from all needles, direct puncture venography was performed, and sclerosant injected with 3% STS foam and bleomycin. Partitioning reduced the overall size of the venous malformation and thus less sclerosant was used. E, An excellent result was achieved after only two treatments.

Box 13-4 Potential Complications Associated With Ethanol and Sodium Tetradecyl Sulphate Sclerotherapy

- Sclerosant effects
 - Local tissue injury (blisters, tissue necrosis, neurologic injury and paralysis, compartment syndrome)
 - Hemoglobinuria
 - Venothromboembolic events
 - Metabolic complications (for example, acidosis, hypoglycemia)*
 - Cardiorespiratory events (respiratory depression, cardiopulmonary collapse, arrhythmias)*
 - Seizures*
 - Perilesional fibrosis (muscle)
- Coagulation derangement
- Procedure plus device risk (from catheters and implantable devices, such as embolic and occlusion devices)
- Inadvertent embolization (of blood clots and embolic agents)
- Age-related risk (such as CNS susceptibility to alcohol in neonates)*

*Sclerosant effects observed with alcohol, whereas all other effects can be seen with both sclerosants.

Major complications are rarely encountered when treating venous malformations; however, sclerotherapy is not without risk. The local risk of tissue injury and compartment syndrome may be seen with STS and ethanol. Occasional closure of large venous channels within the venous malformation or draining the venous malformation can be performed with coils, glue, and endovenous laser ablation. Combining surgical partitioning of a venous malformation with sclerotherapy reduces the amount of sclerosant needed to fill the venous malformation (see Fig. 13-27).

In our practice we have reserved bleomycin for common venous malformations in locations in which postprocedure swelling may be problematic (for example, orbit/eyelids, lips, and tongue, or oral cavity, including the tongue and airway). To reduce washout of bleomycin when injected into venous malformations, a small volume of 3% STS foam is injected immediately before the bleomycin, which results in lesional thrombosis and stasis.

After sclerotherapy, local swelling begins immediately and progresses over several hours (eventually subsiding by 5 to 7 days).⁴¹ Tissue swelling is expected and does not preclude discharge. Pain should be managed appropriately. Significant pain after STS is unexpected. Skin and mucosal blistering is not uncommon when these locations are treated and can be seen before the patient leaves the procedure room or within 24 hours of the injection. Polysporin ointment should be used on skin blisters for 1 week after treatment. Tissue ulceration may occur, although rarely, and should be managed appropriately in conjunction with plastic surgery. Hydration is important after the procedure to minimize the effects of hemoglobinuria, which frequently occurs with STS/ethanol.⁴² Oral fluids are encouraged, along with twice normal IV fluid maintenance given routinely for 4 hours after the procedure. Hemoglobinuria is managed by additional hydration with IV and oral fluids, intravenous furosemide, and occasional urine alkalinization with a sodium bicarbonate intravenous infusion. Measuring urine output and observing urine color during the hospital postprocedural stay is important. All complications require close observation and follow-up⁴³ (Table 13-1).

Most venous malformation sclerotherapy is performed as an elective day-case procedure with a 4- to 6-hour postprocedure stay. Overnight or intensive care admission may be required and arranged as necessary before treatment. In patients with a venous malformation involving an entire extremity requiring sclerotherapy to multiple locations, consecutive daily treatments over 2 to 3 days can be performed if the same location is not treated more than once over the 2 to 3 days. At discharge, the patient should be systemically well, with no concerns regarding the treated site other than the expected swelling and occasional skin blistering.

Common venous malformations have a propensity for recanalization and reenlargement. Therefore repeat injection sclerotherapy treatments are needed in many patients. Staged sclerotherapy is required, and this can be performed, for example, every 6 weeks. Arrangements are made for appropriate clinical and imaging follow-up after treatment.

Table 13-1 Schobinger Clinical Severity Score of Peripheral Arteriovenous Malformations

Stage	Title	Clinical Features
I	Quiescent stage	Warm vascular stain in skin mimicking a capillary malformation
II	Expansion stage	Lesion warmer, bruits and thrills are present, lesion is enlarging
III	Destructive stage	Ulcers, hemorrhage, bone lysis, and pain
IV	Cardiac decompensation stage	Onset of cardiac failure from overload



Fig. 13-28 Outcome after sclerotherapy of labial and lingual venous malformation. After sclerosant injection with small doses of 3% STS foam and bleomycin, good results were seen in these three patients, with no major complications and only mucosal blistering in patient A (clinical follow-up between 3 and 5 years).

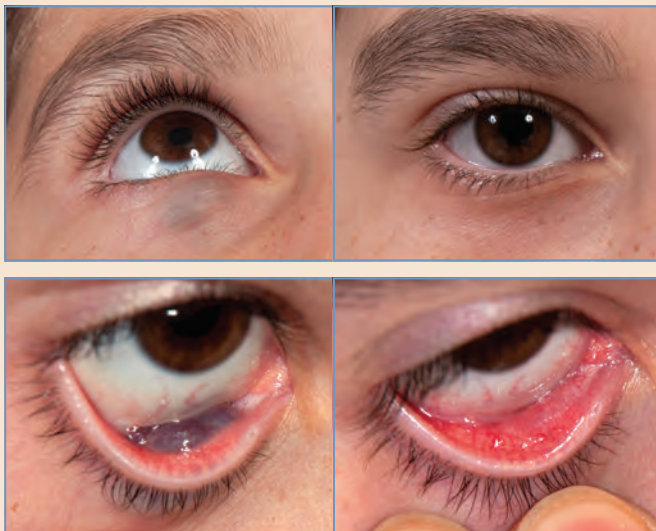


Fig. 13-29 Outcome after sclerotherapy of small eyelid venous malformation. Sclerosant injection with small doses of 3% STS foam and bleomycin resulted in good clinical improvement.

Improvements in pain and disfigurement are expected in most patients undergoing sclerotherapy (Figs. 13-28 and 13-29). Clinical improvements in pain reduction, however, do not always correlate with a reduction of lesion size on imaging. In our practice, we do not perform routine posttreatment imaging unless symptoms persist despite adequate treatment or if further treatment is required for recurrent problems.

Arteriovenous Malformation

Arteriovenous malformations (AVMs) are high-flow vascular malformations, which are very different compared with the more common low-flow vascular malformations.⁴ This section refers to peripheral AVMs and not AVMs involving the central nervous system. An AVM is characterized by an abnormal arteriovenous precapillary shunt, the so-called *nidus*. The nidus differentiates AVMs from congenital AVFs, which are high-flow vascular malformations with precapillary shunts with no nidus. Often multiple arteriovenous shunts are present in AVMs and AVFs. These shunts can be localized or extensive and commonly involve the extremities, trunk, and viscera. AVMs do not respect tissue boundaries, and AV shunting can occur in soft tissues and bone. AVMs usually undergo slow progression and expansion over many years, and during childhood the AVM grows in proportion with the child. The expansion seen in extracranial AVMs is the primary cause of the morbidity associated with these lesions. The Schobinger staging (see Table 13-1) is a clinically based severity scoring system and should be used for all peripheral AVMs (Fig. 13-30).



Fig. 13-30 Clinical features of peripheral AVM. **A**, Schobinger stage I AVM: red vascular skin stain on forehead of an infant. The lesion was warm and pulsatile, and Doppler ultrasonography showed marked hypervascularity with arteriovenous shunts in subcutaneous tissue beneath the stain. **B** and **C**, Schobinger stage II AVM: a warm distal extremity, prominent veins, and palpable thrill from a radial artery aneurysm. **D-F**, Schobinger stage III AVMs: dystrophic skin and mucosal changes from tissue necrosis with ulceration on the pinna and maxillary destruction and bleeding (different patients).

At birth, AVMs appear as a pink cutaneous skin stain and can be mistaken for a capillary malformation or the premonitory sign of an infantile hemangioma (see Fig. 13-30). However, high flow from arteriovenous shunting is present beneath the vascular skin stain, and this can easily be detected with Doppler ultrasonography. The high flow is characterized clinically by increased growth and warmth of the affected body part, bruits, thrills, arterial aneurysms, and enlargement of draining veins (see Fig. 13-30). These features become progressively evident throughout childhood. Arteriovenous shunting can be confirmed by the use of continuous wave Doppler imaging. Tissue ischemia from arteriovenous shunting results in pain and tissue necrosis with ulceration and bleeding (see Fig. 13-30). High-output cardiac failure can be seen in patients with large, extensive AVMs.

Regarding the natural progression of extracranial AVMs, it has been found that as many as 44% of children with Schobinger stage I disease will progress before adolescence and up to 83% before adulthood.⁴³ AVM progression is more likely to occur during adolescence, and diffuse AVMs are more likely to expand in childhood and puberty compared with localized AVMs.⁴³

Local trauma (including iatrogenic trauma) can trigger rapid expansion in these lesions, with increased shunting. Proximal surgical ligation or proximal arterial embolization of feeding arteries will result in the arteriovenous shunts recruiting a vascular supply from adjacent territories. There is still debate whether pregnancy affects AVM progression.

Imaging of Arteriovenous Malformation

AVMs are best imaged noninvasively by MRI, including MRA.⁴⁴ Inflow arteries are enlarged, tortuous, and sometimes aneurysmal. Outflow veins are enlarged and tortuous. Numerous flow voids are evident on spin-echo sequences because of the high flow in arterial channels, and the high density of vessels is easily demonstrated, including the nidi. MRI provides an excellent overall map showing the distribution of the disease and the tissues involved with the shunts. On MRI and ultrasonography, there is no evidence of a parenchymal tumor, which allows differentiation from vascular tumors, some of which have arteriovenous shunts. Soft tissue edema in AVMs can be seen on MRI surrounding the arteriovenous shunts.

It is important to assess the surrounding soft tissue around an AVM. AVMs located in large fatty areas can occur in *PTEN* gene mutation–related AVMs (for example, in *Banyan-Riley-Ruvalcaba syndrome*). However, details of feeding arteries, shunt morphology, and venous drainage are better assessed by catheter angiography and direct puncture techniques (Fig. 13-31).

Catheter angiography remains the benchmark procedure in imaging; however, it is not required until treatment is needed and is usually performed at the time of the first embolization. For example, the first catheter angiogram on a patient with a peripheral AVM should include “global” and “selective” angiography (see Figs. 13-9 and 13-31). Diagnostic angiography should always try to demonstrate the following:

- Arterial runoff throughout the affected extremity
- Feeding arteries to shunts
- Location of the shunts
- Shunt morphology
- Venous drainage from the shunts (see Figs. 13-9 and 13-31)

Carefully studying the angioarchitecture can help to plan endovascular treatment.⁴⁵

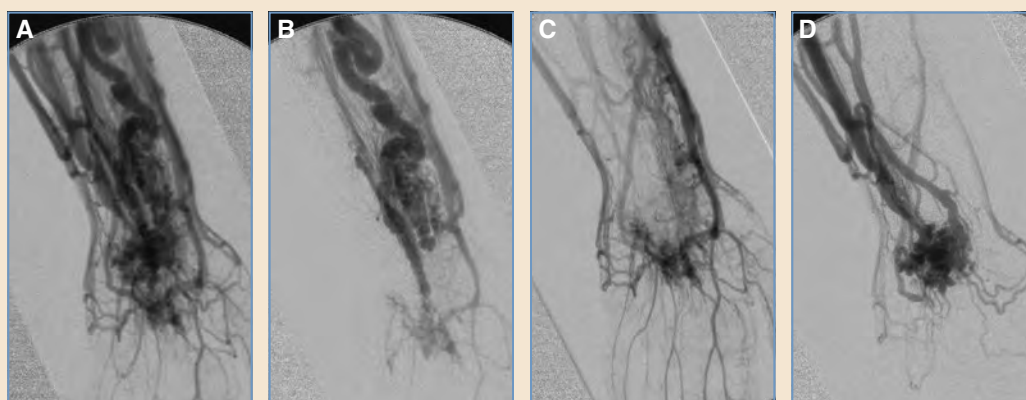


Fig. 13-31 Catheter angiography demonstrating arteriovenous shunts (nidi) in an upper extremity AVM. **A**, Global catheter angiography can show arteriovenous shunts/nidi in the forearm and hand. Detailed angiography is needed to fully evaluate arteriovenous shunts by selective catheterization of the interosseous, radial, and ulnar arteries. **B**, Interosseus artery. **C**, Radial artery. **D**, Ulnar artery.

Treatment of Arteriovenous Malformation

Many patients are observed with AVMs in the early stages and are monitored carefully throughout childhood. Most AVMs will eventually require treatment because of continued lesion expansion. Lesion expansion can be associated with troublesome symptoms, including tissue ischemia and ulceration, pain, and sometimes cardiac overload.⁴ Although the ultimate treatment goal is complete shunt occlusion and surgical resection, this goal cannot always be achieved.

The mainstay of treatment to date has been endovascular treatments with catheter embolization and sometimes surgery. Endovascular treatments with catheter embolization can at best provide control and do not cure the AVM. Although embolization, either alone or in combination with surgical excision, has been the primary treatment for many, it has recently been suggested that well-localized stage I AVMs that are amenable to surgical excision have better outcomes without preoperative embolization.^{43,46,47} However, the full extent of the AVM may be underrecognized during early disease; therefore embolization and surgery during infancy and early childhood are rarely performed because of the high risk of disease progression. Embolization and resection produce a proangiogenic environment, resulting in lesion enlargement. Disease recurrence is high after treatment and is reported in up to 93% of cases.⁴³ This propensity for progression or recurrence after certain treatments poses many challenges, which are compounded by the high risk from treatment.⁴⁶⁻⁴⁹ There is no “quick fix” for treating AVMs, and most require repeated treatment sessions over several years. When surgical resection is performed after embolization, surgery should be performed within a few days of the embolization. Embolization can decrease intraoperative bleeding; however, it does not reduce the amount or extent of tissue that must be resected. Embolization alone can be useful for symptom control in patients with extensive disease when no other suitable treatment options are available (Fig. 13-32). Surgical amputation may be considered for advanced extremity AVMs that cause major problems threatening the affected limb or the patient’s life (see Fig. 13-32).

Cures are rarely seen, and all patients require long-term follow-up after treatment. Interestingly, AVMs that fail to show reexpansion for 5 or more years after intervention are more likely to have long-term control.⁴³

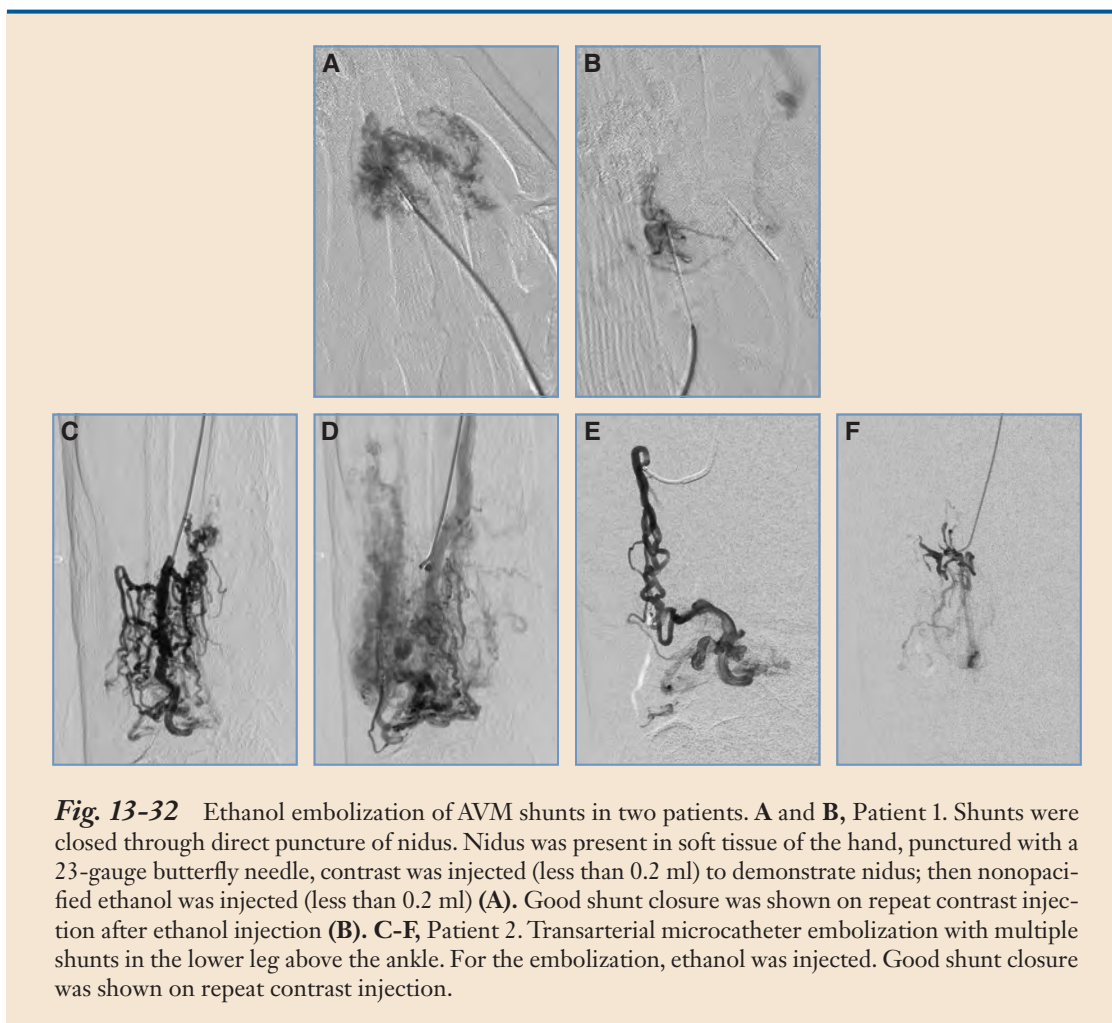


Fig. 13-32 Ethanol embolization of AVM shunts in two patients. **A** and **B**, Patient 1. Shunts were closed through direct puncture of nidus. Nidus was present in soft tissue of the hand, punctured with a 23-gauge butterfly needle, contrast was injected (less than 0.2 ml) to demonstrate nidus; then nonopacified ethanol was injected (less than 0.2 ml) (**A**). Good shunt closure was shown on repeat contrast injection after ethanol injection (**B**). **C-F**, Patient 2. Transarterial microcatheter embolization with multiple shunts in the lower leg above the ankle. For the embolization, ethanol was injected. Good shunt closure was shown on repeat contrast injection.

Embolization of Arteriovenous Malformation

No routine preprocedural workup is needed for the embolization of AVMs. However, the anesthesiologist must be aware of cardiac decompensation. The aim of endovascular treatment of AVMs is to selectively close the arteriovenous shunts. These are all complex procedures, and nontarget embolization should be avoided, because this can lead to significant tissue complications, including neurologic sequelae with paralysis.

Shunt closure can be achieved through one or more of the following techniques (see Fig. 13-32; Figs. 13-33 and 13-34):

- Direct percutaneous puncture of the AVM nidus (for nonvisceral AVMs)
- Transarterial catheter embolization using a coaxial catheter system with a microcatheter advanced as close as possible to the nidus
- Embolization of the venous outflow from the nidus,^{50,51} either by direct puncture of the vein outflow or by retrograde venous catheterization

The endovascular treatment strategy is determined by the patient's symptoms. In those patients in whom venous congestion or hemorrhage is particularly troublesome, closure from the arte-

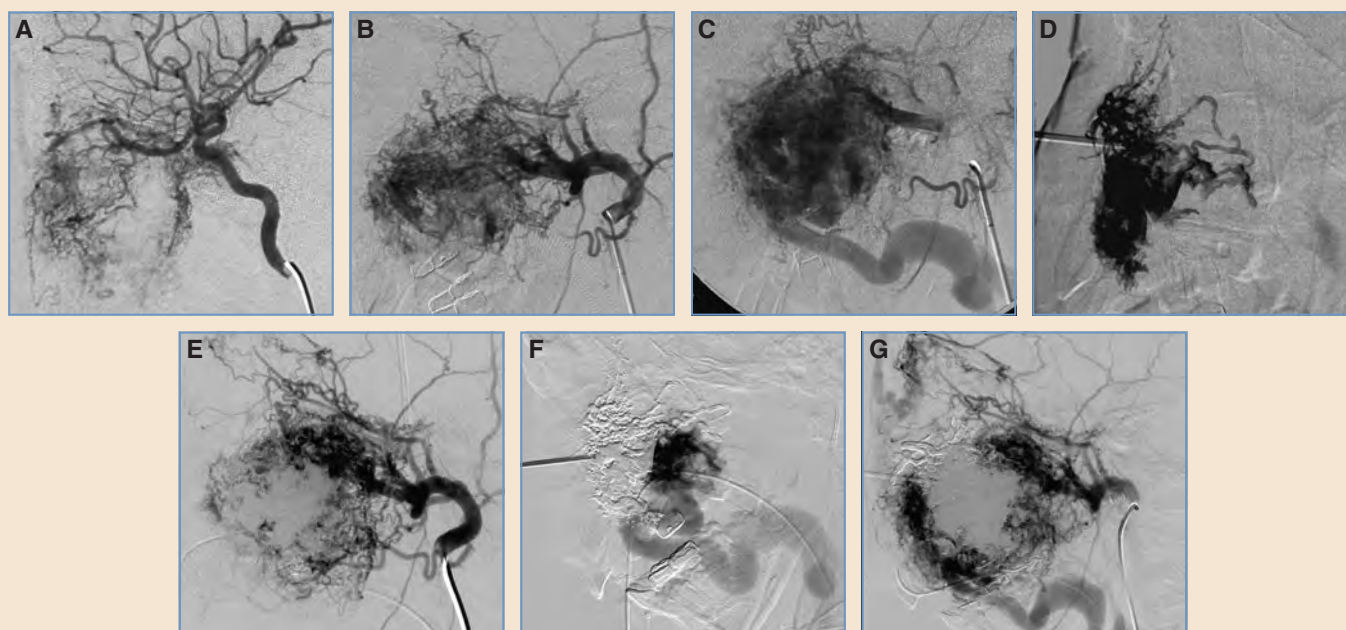


Fig. 13-33 Onyx embolization of maxillary AVM (same patient as in Fig. 13-30, *E* and *F*). **A**, Schobinger stage III AVM supplied bilaterally by internal carotid arteries (descending ophthalmic artery branches). **B**, External carotid arteries (internal maxillary artery branches). **C**, Prompt venous drainage. **D**, Direct puncture (18-gauge needle) into the maxillary nidus. **E-G**, Subsequent Onyx injections as serial procedures over 2 years, showing progressive closure of much of the arteriovenous shunts.

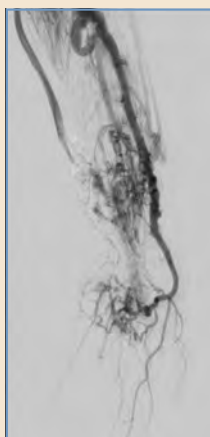


Fig. 13-34 Angiographic result of AVM embolization (same patient as in Fig. 13-31). Serial embolizations with ethanol and glue were performed to control pain; both agents were injected by direct puncture and transarterial techniques at different shunt locations. Good symptom control was achieved. Completion angiogram at symptom control shows significant closure to multiple shunts in the distal forearm and hand, with preservation of digital flow. Distal ulnar artery occlusion occurred from embolizations.

rial side with an arterial catheter is preferred before closure of the venous end of the shunt. In patients with tissue necrosis, embolization from the venous end should be considered before using an arterial approach.

As AVMs invariably have many nidi, the question often asked is which nidus/nidi should be closed? With diffuse extremity disease, nidal closure at the location of symptoms such as pain would be appropriate. In patients with cardiac overload, closure of the largest shunts initially would be appropriate (Fig. 13-35).



Fig. 13-35 Recommended treatment options for extremity AVMs. **A** and **B**, Recurrent bleeding from focal thumb AVM (thumb functional)—initially embolization and if needed amputation of digit. **C** and **D**, Pain from diffuse AVM of hand (hand functional)—for embolization, no surgical option. **E**, Chronic ulceration—for amputation. **F-I**, Diffuse truncal and extremity AVM, multiple life-threatening hemorrhages—palliative embolizations performed, no surgical option.

Embolic agents used to close arteriovenous shunts include liquid embolics and coils. All agents need to be safely delivered to the target. A coaxial catheter system is essential when agents are delivered transarterially. A coaxial system uses an outer guiding catheter through which the “delivery” microcatheter for the embolics is passed. The outer catheter provides stability for the microcatheter to reach its target and also allows multiple changes of microcatheters. The commonly used liquid embolics are ethanol and the adhesive glue, N-butyl-2-cyanoacrylate (Histoacryl). The nonadhesive liquid embolic ethylene vinyl alcohol copolymer (Onyx) is favored by some interventional radiologists. Despite being the most destructive vascular endothelial agent, ethanol is not favored by some because of its associated life-threatening intraprocedural cardiopulmonary complications. However, when used appropriately, the angiographic results from the use of ethanol can be excellent. Histoacryl glue and Onyx can result in satisfactory angiographic shunt closure; however, they do not destroy the vascular endothelium, as does ethanol. Any of the three liquid agents can be injected through a microcatheter or direct percutaneous methods. Ethanol must never be injected into visceral AVMs, because the risk of parenchymal necrosis is high. Histoacryl glue is a useful alternative for visceral AVMs. In pediatrics, success has been reported for novel lifesaving neonatal liver AVM embolizations with excellent long-term outcomes.^{52,53} Traditionally, coil embolization is not recommended in AVMs; however, this technique can be useful to close the venous end of selected arteriovenous shunts, because this promotes closure of the shunts themselves.^{48,49} Flow-related aneurysms are well recognized and can be problematic and challenging to treat (Fig. 13-36).

After embolization, local swelling as expected starts immediately, and progresses over several hours, eventually subsiding by 5 to 7 days. Pain should be managed appropriately. Local tissue injury with necrosis can occur and affect skin and mucosa, with early signs evident within 24 hours; it requires careful follow-up and appropriate care in conjunction with plastic surgery. Hydration is important after the procedure to minimize the effects of hemoglobinuria, which frequently occurs with ethanol.³⁴ Oral fluids are encouraged, along with twice normal IV fluid maintenance given routinely for 4 hours after the procedure. Hemoglobinuria is managed by additional hydration with IV and oral fluids, intravenous furosemide, and occasional urine alkalinization with a sodium bicarbonate intravenous infusion.³⁴ Bed rest for approximately 6 hours is recommended when arterial catheterization is performed through the femoral artery. All relevant peripheral pulses are monitored during the first 6 hours after the procedure. All complications (see Box 13-4) require close observation and follow-up.⁴⁶⁻⁴⁹

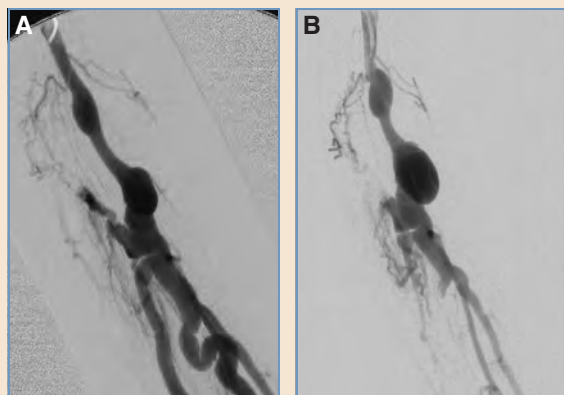


Fig. 13-36 Flow-related aneurysms in peripheral AVMs. These occur on the arterial inflow to the arteriovenous shunts. Progressive increase in size over the years can occur. **A**, Aneurysm in patient at 12 years of age. **B**, In the same patient at 17 years of age.

AVM embolization is usually performed as an elective procedure with a 24-hour postprocedural stay. Elective intensive care admission after the procedure is needed if the AVM involves vital areas, such as the airway or oral cavity, including the tongue. At discharge, the patient should be systemically well, with no concerns regarding the treated area other than expected swelling.

Repeated embolization procedures are typically required because of the large number of shunts and a propensity for the shunts to open after treatment is started. Repeat AVM embolization is performed approximately every 8 weeks. Arrangements are made for appropriate clinical and imaging follow-up after treatment. Patients often require multiple angiographic catheterizations. The high complication risk associated with AVM embolization is the result of local and systemic problems related to the injected embolic agents, catheter technique, and devices used and risks from inadvertent embolization (see Table 13-1). Despite this, patients can often have significant improvement in their symptoms with embolization.

VASCULAR MALFORMATIONS ASSOCIATED WITH OTHER ANOMALIES

Vascular malformations (simple and/or of major named vessels) may be associated with anomalies of bone, soft tissue, or viscera. These nonvascular anomalies are often soft tissue and are sometimes associated with skeletal overgrowth or rarely undergrowth. In patients with PTEN gene-mutation-related overgrowth and vascular malformations, lifelong screening for associated malignancies is required throughout adulthood. The eponymous syndromes used for these anomalies can cause confusion, and it is always important to describe the individual vascular channel malformations and associated anomaly. In selected patients, the interventional radiologist can help with these complex disorders, but there are no cures.

Klippel-Trenaunay Syndrome

KTS is a low-flow combined vascular malformation involving abnormal capillary, lymphatic, and venous channels associated with prominent soft tissue and bony hypertrophy.^{54,55} Commonly, the malformation affects a single lower extremity. However, more than one extremity can be affected, usually asymmetrically. Macrodactyly can be seen in an uninvolved limb. The trunk (thorax, perineum, and retroperitoneum) can also be affected. The original description of KTS was a triad of physical findings: a cutaneous capillary malformation (port-wine stain), varicosities, and hypertrophy of soft tissue and bone in the affected region (Fig. 13-37). Like many vascular anomalies, the severity of the phenotype and component anomalies in KTS is variable.

Lymphatic malformations are also seen in KTS, in addition to other low-flow malformations, in the original description. This syndrome is sporadic and evident at birth. The capillary malformation component is present in the skin often as a “geographic” stain. The venous component consists of anomalous superficial embryonic veins, such as the lateral marginal vein of Servelle⁵⁶ (see Fig. 13-7). Deep venous anomalies can occur, such as hypoplasia and segmental atresias (Fig. 13-38). In lower extremity disease, venous hypertension occurs and varicosities may worsen with increasing age. The lymphatic component (lymphatic malformation) is variable and includes conducting lymphatic channel hypoplasia, lymphedema, and cystic lymphatic malformations. Microcystic lymphatic malformations are often located on the surface of the cutaneous capillary malformation and can bleed intermittently.

Extremity overgrowth is obvious at birth, and although progressive, major changes after birth are unusual. The overgrowth of an affected limb can involve the entire extremity, including the hand and foot. When a leg-length malformation affects the lower extremity, yearly monitoring is needed, because a leg-length discrepancy can occur. Radiographic monitoring of leg length

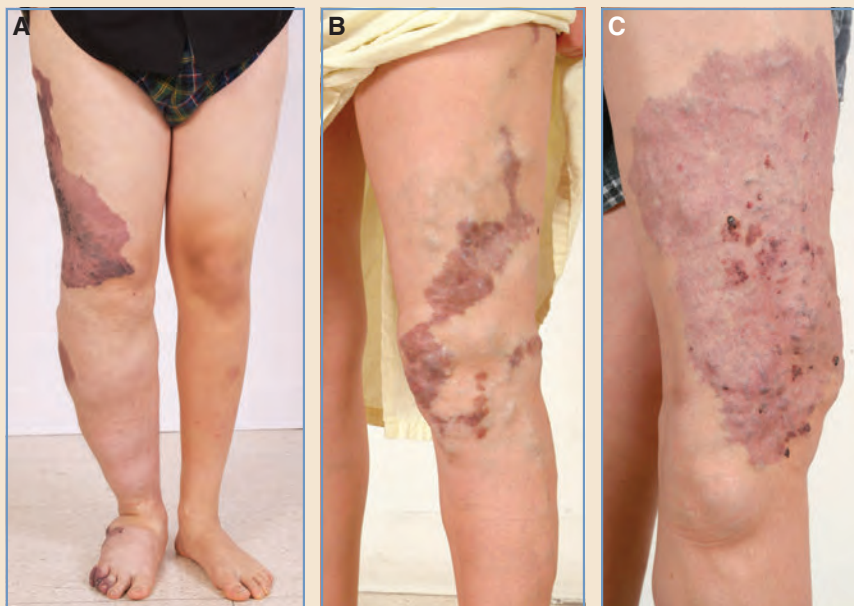


Fig. 13-37 Clinical features of KTS. Typical physical findings in three patients with lower extremity KTS with extremity overgrowth. **A**, Capillary malformation (port-wine stain), lymphatic malformation on surface of stain as small dark vesicles. **B** and **C**, Capillary malformation (port-wine stain), lymphatic malformation on surface of stain as small dark vesicles, and lateral viscosities—venous malformation.

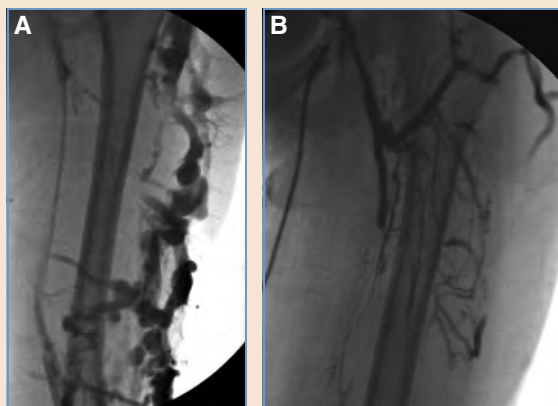


Fig. 13-38 The deep venous system in lower extremity KTS. Venograms in two patients showing deep vein anomalies. **A**, Femoral vein hypoplasia. A venogram is performed through pedal vein injection with an ankle tourniquet to divert flow into the deep veins, also showing a multichannel embryonic lateral marginal vein. **B**, Femoral vein atresia. A venogram is performed through catheter injection into the common femoral vein with groin compression to promote retrograde filling.

is performed in those children with a leg-length discrepancy who are older than the toddler age group to assess the need for timed surgical correction with an epiphysiodesis. Infrequently, there can be undergrowth of the affected limb. The affected limb may also be enlarged in girth. However, this is the result of a combination of thickened skin, excessive fat, and an increase in abnormal vascular tissue. The muscles are usually normal in size and sometimes smaller. Vascular anomalies can be present in the muscle compartments.

Extremity pain is not uncommon and can be from various causes, such as venous hypertension and chronic venous insufficiency, cellulitis, deep venous thrombosis, thrombophlebitis, osseous venous malformations, arthropathy, neuropathy, and growing pain.⁵⁷ Thrombophlebitis of the anomalous veins occurs in up to 45% of patients, and pulmonary emboli are reported in between 4% and 25% of patients. Pulmonary hypertension has been reported in patients with KTS, possibly caused by silent recurrent pulmonary emboli.³⁴ Bleeding can be problematic from the skin and the genitourinary and gastrointestinal tracts. Patients can also have recurrent lymphatic malformation infections, cellulitis, and lymphedema.

Klippel-Trenaunay Syndrome Imaging and Treatment

Imaging plays an important role in the evaluation of KTS patients.⁵⁸ Radiographs (including CT scanography/digital radiography) are performed to measure length discrepancies in the lower extremities. MRI is the best imaging modality to study the soft tissue components of the affected area and easily demonstrates the abnormalities in the subcutaneous tissues (lymphatic malformations, venous malformations, anomalous veins, and increased fat) and muscle compartments, which may appear enlarged because of infiltration by lymphatic and venous malformations.⁵⁸ Magnetic resonance venography (MRV) can help to define the anatomy of the deep venous system and venous anomalies. The deep venous system may be hypoplastic with segmental atresias and therefore difficult to see on MRV. Further invasive assessment with conventional venography is performed when endovascular closure of venous anomalies and embryonic veins is planned. Invasive venography can be performed through a variety of techniques, including conventional

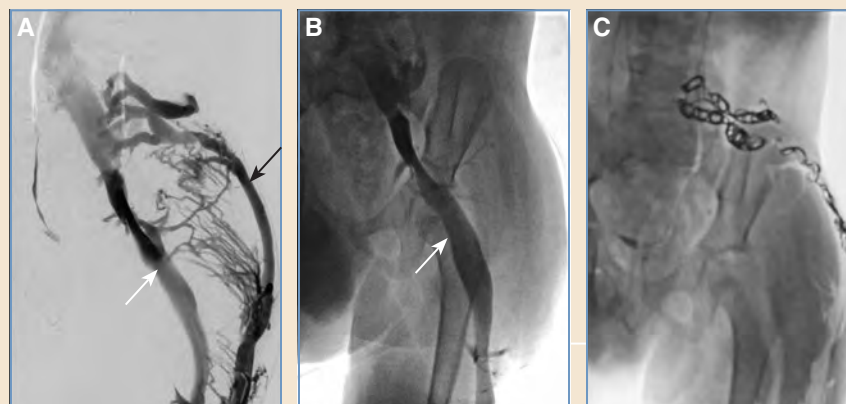


Fig. 13-39 Endovascular closure of embryonic vein in KTS. **A** and **B**, Venography shows both embryonic veins present: the lateral marginal vein (*black arrow*) draining into the common iliac vein, and the sciatic vein (*white arrow*) draining into the internal iliac vein. **C**, Endovascular closure of the lateral marginal vein performed by coil embolization of the intrafascial more deeply located in the upper end of the vein communicating with the common iliac vein and then endovenous laser ablation to the more superficial extrafascial lower end of the vein.

pedal venography, percutaneous injection of tibial veins, retrograde catheter venography, and diversion venography⁵⁹ (see Figs. 13-8 and 13-38; Fig. 13-39). Contrast venography is challenging because of the large capacity of the anomalous channels and hypoplasia of the deep veins. In the young patient, the embryonic lateral marginal vein is typically large but straight and becomes more tortuous in the older patient. This embryonic vein may be either a single channel or multiple interconnecting longitudinal channels (see Figs. 13-38 and 13-39).

For most patients, treatment is conservative.^{54,55} When lower extremities are affected, a leg length discrepancy is managed appropriately. Specific problems must be addressed when they arise, such as infections, bleeds, thromboembolic events, and extremity pain. Graded compression stockings can be extremely helpful in those patients with axial limb enlargement. Patients comment that their affected extremity can feel “lighter,” and in particular, compression garments can help with the subcutaneous venous anomalies, venous hypertension, chronic venous insufficiency, and lymphedema. A variety of topical treatments can be effective to treat skin bleeding.

Sclerotherapy can be offered to treat certain components, such as focal venous malformations, including perirectal venous malformations causing rectal bleeding, macrocytic lymphatic malformations, and bleeding cutaneous lymphatic vesicles. Recurrence after sclerotherapy, however, is recognized. Closure of the embryonic veins may offer some protection against venothromboembolic disease in KTS, and this can be achieved either through an endovascular approach using endovenous laser and coil embolization (see Fig. 13-39) or sometimes surgically (Fig. 13-40). Treatment for venous anomalies may be required when these become symptomatic, and either an endovascular or surgical approach can be performed⁶⁰⁻⁶³ (see Figs. 13-39 and 13-40). Secondary procedures are required in a significant number of patients after surgical treatments of the venous anomalies. In those patients with massive overgrowth of soft tissue, staged surgical debulking of the extrafascial compartment can be offered.⁶⁴

Prophylactic anticoagulation should be considered for KTS patients when they are undergoing long and complex interventions and surgery because of increased risk of venous thromboemboli.

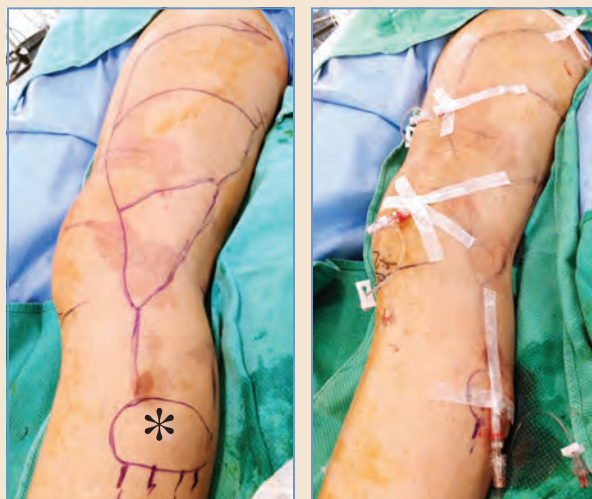


Fig. 13-40 Mapping the extrafascial embryonic lateral marginal vein, its tributaries, and superficial varix for endovascular laser closure. The location of these channels, including the varix (*), are marked on the skin to access them appropriately for endovenous closure.

Parkes Weber Syndrome

Parkes Weber syndrome is a complex, combined high-flow vascular malformation with extensive capillary staining, diffuse arteriovenous shunts, and rarely lymphatic malformations. The lymphatic malformations include cutaneous vesicles, lymphedema, dilated conducting lymphatics, and chylous reflux into the limb. Macrocystic lymphatic malformations are not seen in Parkes Weber syndrome.^{58,62} The clinical findings of high flow (from the arteriovenous shunts) are characterized by localized hyperthermia, bruits, thrills, and varicosities. The condition is obvious at birth and appears as overgrowth with a “geographic” macular pink skin stain. The stain is variable in size and appearance; for example, some are faint in color. Skin, subcutaneous tissues, and muscle are affected. The extremity overgrowth is axial and longitudinal, and the disease may have a focal distribution or involves an extremity in a more diffuse/multifocal pattern (Fig. 13-41). The limb overgrowth affects soft tissue (including muscle) and bone. Arteriovenous shunting occurs through macroarteriovenous and microarteriovenous fistulas, and varicosities result from arteriovenous shunting (Figs. 13-42 and 13-43). The vascular skin stain is not a true capillary malformation as in KTS and has been described as a “pseudocapillary malformation,” because it is most likely caused by microarteriovenous fistulas, which explains the warmth when the stain is palpated, unlike the capillary malformation in KTS.

Physical findings of Parkes Weber syndrome can be confirmed easily with continuous wave Doppler examination, which shows arteriovenous shunting. Arteriovenous shunting may lead to cardiac decompensation from volume overload and can present at birth or later (see Fig. 13-43). Parkes Weber syndrome has been confused with KTS because of the extremity skin stain, varicosities, and overgrowth; in contrast, KTS has no high-flow component and does not affect extremities in a focal distribution. Macrocystic lymphatic malformations are not seen in Parkes Weber syndrome but can occur in KTS. Therefore it is easy to differentiate Parkes Weber syndrome from KTS. Although Parkes Weber syndrome can be sporadic, it belongs to the clinical spectrum of capillary malformation–arteriovenous malformation, and approximately 50% of Parkes Weber syndrome cases are associated with a *RASA1* gene mutation.⁶⁵



Fig. 13-41 Clinical features of Parkes Weber Syndrome, as shown in four patients. **A** and **B**, Patient 1. Pedal overgrowth with multifocal pink geographic macular skin stains and prominent subcutaneous veins in the distal lower extremity.

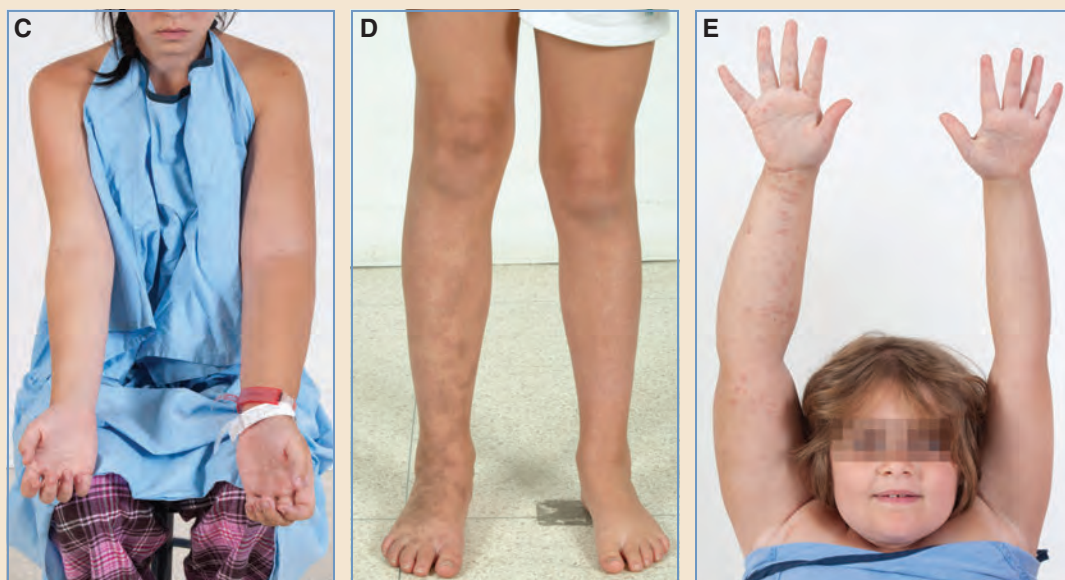


Fig. 13-41, cont'd C, Patient 2. Teenage girl with cardiac decompensation and a very faint macular pink skin stain and generalized overgrowth of a “warm” left upper extremity. D, Patient 3. Right lower extremity overgrowth (especially longitudinal) in 7-year-old girl with a warm lower extremity and multifocal dark macular skin stains. E, Patient 4. Generalized overgrowth of the right upper extremity in patient with diffuse “spotty” macular skin stain and a warm affected extremity.

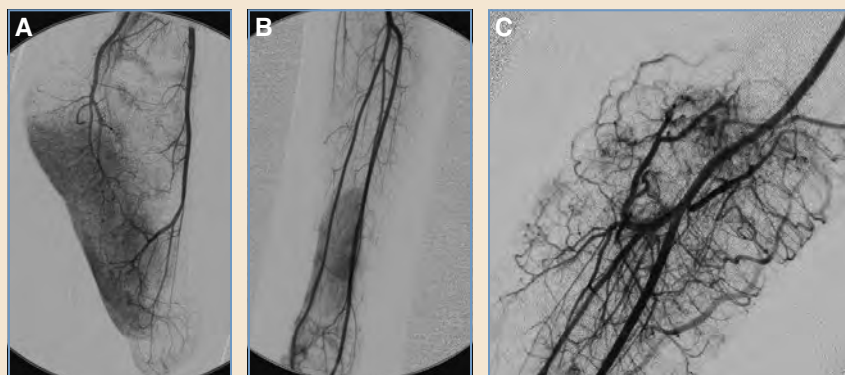


Fig. 13-42 Parkes Weber syndrome microarteriovenous fistulas on catheter angiography. Two patients with angiographically different microarteriovenous shunts. A and B, Patient 1 (Patient 1 in Fig. 13-41) shows soft tissue staining from microarteriovenous shunts in the foot and calf, with no direct arteriovenous shunts seen. C, Patient 2 (Patient 4 in Fig. 13-41) shows diffuse circumferential microarteriovenous shunts with microarteriovenous fistulas seen. Both patients are unsuitable for catheter embolization, because this cannot be performed for microarteriovenous fistulas without significant complications.

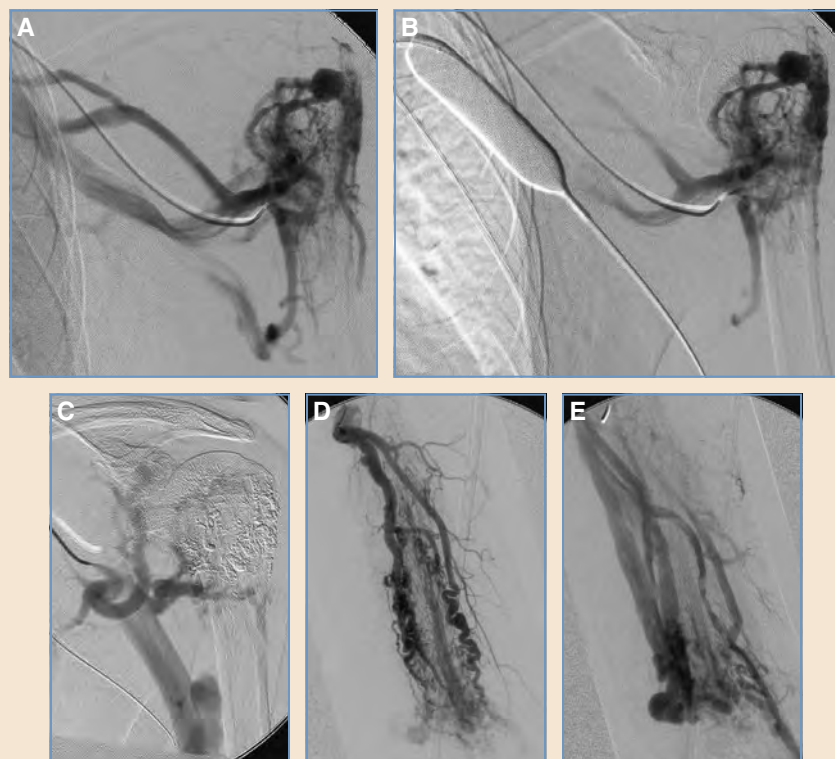


Fig. 13-43 Angiography and embolization of macroarteriovenous fistulas in Parkes Weber syndrome (Patient 2 in Fig. 13-41). **A**, Patient with cardiac decompensation has multiple macroarteriovenous shunts in the upper arm fed by the circumflex humeral artery. **B**, During embolization of circumflex humeral shunts, venous outflow was controlled using an endovascular balloon in the subclavian vein. **C**, Macroarteriovenous shunts embolized successfully with glue using a coaxial microcatheter technique. **D** and **E**, Multiple macroarteriovenous shunts in the upper arm fed by the profunda brachii arteries. Staged embolization of multiple macroarteriovenous fistulas in the left upper extremity resulted in significant improvement of cardiac status.

Parkes Weber Syndrome Imaging and Treatment

MRI is recommended to evaluate the overall distribution of Parkes Weber syndrome and demonstrate the subcutaneous extrafascial anomalies, including abnormal vascular tissue, microcystic lymphatic malformations, lymphedema, and fatty overgrowth.⁵⁸ Unlike in KTS, muscle and bone hypertrophy is seen in Parkes Weber syndrome. In an affected extremity, enlarged inflow arteries and outflow veins are seen and numerous flow voids are present on spin-echo sequences. Microarteriovenous shunts can be seen as T2 signal hyperintensity in deeper tissues such as muscle.

Bony hypertrophy can result in a limb-length discrepancy, and epiphysiodesis may be required. Surgical debulking is not generally performed, because overgrowth affects muscle and bone.

Catheter angiography can demonstrate microarteriovenous and macroarteriovenous shunts (see Figs. 13-42 and 13-43). Catheter angiography is not usually performed unless the patient has local pain, tissue ulceration, or cardiac decompensation. The catheter angiogram can identify macroarteriovenous fistulas, which may be suitable for endovascular closure, resulting in symptom improvement. Diffuse microarteriovenous shunts can be seen in the subcutaneous fat and

muscle and are unsuitable for catheter embolization. The AVFs may occur throughout an entire extremity or more focally. Endovascular closure of macroarteriovenous fistulas reduces shunting, leading to improved tissue perfusion with reduced pain, healing of skin ulceration, and control of cardiac overload (see Fig. 13-43).

Both KTS and Parkes Weber syndrome are complex vascular malformations, and although there are no cures, the interventional radiologist can provide useful therapeutic options with symptom improvement.

KEY POINTS

- Patients with vascular anomalies will benefit from a multidisciplinary team approach to patient care.
- Two features that help differentiate vascular tumors from vascular malformations are (1) the patient's age when the lesion was first noticed and (2) the growth of the lesion over time and if the growth is commensurate with that of the child.
- The postnatal diagnosis of a vascular anomaly can be made in most patients based on history and physical examination.
- The antenatal diagnosis of vascular anomalies relies on imaging with fetal ultrasonography and MRI.
- Vascular malformations progress throughout childhood and persist throughout adulthood. Low-flow malformations can be differentiated from high-flow malformations on physical examination.
- The two most helpful imaging modalities used to establish or confirm a diagnosis and define disease extent are ultrasonography and MRI.
- Imaging plays a significant role in the diagnosis of vascular anomalies when lesions are deep with no visible cutaneous signs.
- Basic MRI with T1, T2, and postcontrast sequences (sometimes with fat saturation) is highly tissue specific when imaging vascular anomalies. MRA sequences are not required to establish a diagnosis.
- "Bedside ultrasound" is extremely useful in a clinic setting to provide and/or confirm a diagnosis in selected patients. A high level of skill is required to perform and interpret the ultrasonography.
- Most vascular anomalies are not life-threatening; however, some can threaten life because of associated severe coagulopathies and hemorrhage, cardiac failure, venothromboembolic events (including pulmonary embolism), sepsis, pressure effects on vital structures, associated malignancies, and iatrogenic insults/inappropriate therapies.

Continued

KEY POINTS (continued)

- Not all vascular anomalies require treatment. The need for treatment is often determined by the lesion and its associated effects.
- Medical therapies are often used for vascular tumors and radiologic/surgical therapies for vascular malformations. Multimodal therapies may be required in some cases.
- Infantile hemangioma is the most common tumor of infancy and along with all vascular tumors should be managed by a pediatric dermatologist.
- Most babies with infantile hemangiomas do not require imaging.
- Capillary malformation (port-wine stain) is the most common vascular malformation. Treatment, if required, is performed with pulsed dye laser.
- Cystic lymphatic malformations can be differentiated from common venous malformations, because they are not fully compressible, show no refilling on dependency/increased venous pressure, and do not have phleboliths.
- It is important to image a cystic lymphatic malformation before treatment to assess the amount of macrocystic and microcystic components within the lymphatic malformation. This aids in appropriate treatment planning.
- Sudden expansion is unique to cystic lymphatic malformations and occurs as a result of self-limiting intralesional hemorrhage or infection.
- In patients with venous malformations, high D-dimer and low fibrinogen levels indicate a localized intralesional coagulopathy within the venous malformation. D-dimer elevation in this setting does not indicate a deep venous thrombosis in most patients.
- The coagulopathic risk in low-flow malformations occurs in two main groups of patients: those with diffuse venous malformations in which there is a localized intralesional coagulopathy, and those with embryonic veins, typically in KTS and CLOVES syndrome.
- Radiologic treatments are an important first-line therapy for many patients with cystic lymphatic malformations, common venous malformations, and AVMs. Many patients can be helped with sclerotherapy, embolization, and laser techniques.
- AVMs are some of the most challenging vascular malformations, with lesions controlled rather than cured by present-day therapeutic approaches. High recurrence rates are seen in most patients treated for extracranial AVMs.

- KTS is a combined low-flow malformation with overgrowth. The management of KTS is mainly conservative. Prophylaxis against venothromboembolic events, during high-risk interventions and avoidance of other thrombotic risk factors (including the use of the oral contraceptive pill), must be considered.
- Parkes Weber syndrome is a combined high-flow malformation with overgrowth. If required, endovascular catheter closure (embolization) can be performed if the arteriovenous shunts are large enough. The microarteriovenous fistulas commonly seen cannot be treated by embolization.

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14

Conjoined Twins

Richard J. Redett • Ronald M. Zuker

HISTORY

Conjoined twins have fascinated the medical profession and general public since antiquity. Early Greek and Roman scholars theorized that maternal impressions or mortal imperfection and parental sin were to blame for the birth of conjoined twins.¹⁻³ Reflecting the social prejudices of the era, conjoined twins were often referred to as monsters. These children were felt to be warnings from God to the parents to amend their life against immorality.¹ Conjoined twins who survived were religious and societal outcasts and were often forbidden burial in consecrated ground.

In his textbook, *De Generatione Animalium* (*On the Generation of Animals*), Aristotle attributed conjoined twins to the quantity of semen and the shape of the womb. Wombs that were too small constrained the seed of twins, causing them to become congested and joined. His theories of embryogenesis prevailed well into the thirteenth century.

Sixteenth-century surgeon Ambroise Paré⁴ reported that Hippocrates believed “too great a quantity of seed” was responsible for multiple births and conjoined twinning. In contrast, “a seed that is lacking in quantity” was thought to lead to limb agenesis and missing body parts. In his treatise, *On Monsters and Marvels*, Paré⁴ described several cases of imaginary and real parapagus, craniopagus, ischiopagus, and parasitic conjoined twins. He also provided illustrations from twin specimens he reportedly had in his possession.

The Biddenden Maids, Mary and Eliza Chulkhurst, are probably the first documented case of conjoined twins.⁵ Born in the village of Biddenden, Kent, England in 1100, they spent their lives joined as pygopagus twins. Early illustrations depicted the sisters united at the hips and shoulder (Fig. 14-1, *A*), although they were instead probably joined only at the buttocks. They lived a prosperous existence and at their death, bequeathed a modest plot of land to the village parish.

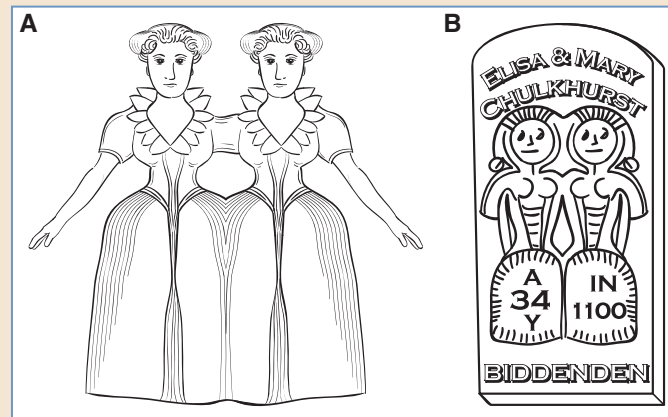


Fig. 14-1 A, Pygopagus twins—the Biddenden Maids. B, Biddenden Maid cakes were distributed annually to the poor in memory of the sisters.

Interest on the annual rental of the land was used to distribute cakes bearing the sisters' image to the poor on Easter Sunday (Fig. 14-1, B). After the death of one twin at age 34, the other sister refused to be separated, stating, "As we came together, we will go together." The second sister died 6 hours later.

Dicephalus (now called *parapagus*) conjoined twins have attracted much attention. Historical illustrations depict these individuals as sharing a single body with two separate heads. The earliest authenticated case is of two Scottish brothers born in 1490.⁶ With the help of King James IV of Scotland, these children became talented musicians and linguists. They lived to the age of 28.

The most well-known parapagus twins were the brothers Giovanni and Giacomo Tocci, born in northern Italy in 1877⁶ (Fig. 14-2). Their social and medical history is well documented, providing much insight into their lives.^{6,7} When the children were 1 month old, they were put on exhibit at the Turin Academy of Medicine for examination by physicians and the public.⁷ They shared a pair of normal legs and were joined to the level of the sixth rib. Each twin had independent control of one leg, making coordinated walking difficult. This separate control of each limb has interesting implications, as will be discussed later in this chapter. As with most "double monsters," they unwillingly led their life touring the world, being promoted with broadsides and pamphlets as the "Two-Headed Boy," and the "Greatest Human Phenomenon Ever Seen Alive." Tired of the degrading nature of living as a sideshow attraction, the twins retired into secrecy at the age of 20.

The original Siamese twins, Cheng and Eng Bunker, were medical and public wonders of the nineteenth century (Fig. 14-3). Connected by a thin abdominal band (omphalopagus), they came to public attention as a display in P.T. Barnum's circus. They married different women (who were sisters), fathered 21 children, and went on to become successful businessmen and farmers. They frequently argued and appealed to their physician for separation. Their request was always denied, citing sure death if surgery were ever undertaken. Autopsy after their death demonstrated a small "tract of portal continuity" consisting primarily of skin, hepatic, and vascular tissue that would have made separation relatively easy today—and then.⁸

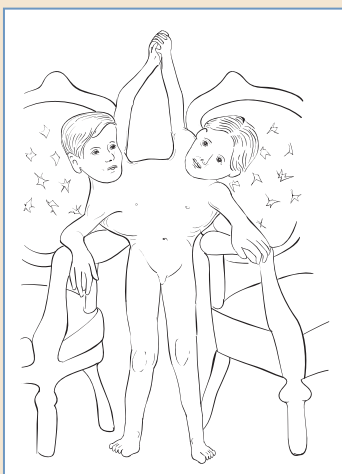


Fig. 14-2 Parapagus twins—the Tocci brothers from Northern Italy.

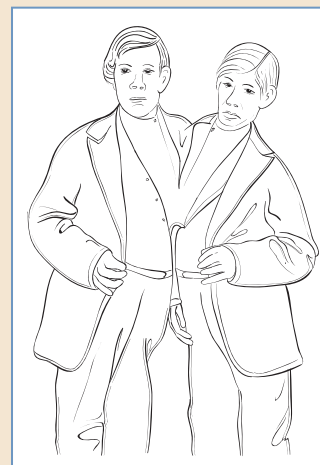


Fig. 14-3 The original Siamese twins, Cheng and Eng Bunker.

CAUSES

Until recently, incomplete fission of a fertilized ovum was the accepted hypothesis for the cause of conjoined twins.⁹⁻¹¹ New insight into embryogenesis has provided evidence that conjoined twins “result from the secondary union of two originally separate monovular embryonic discs.”¹² All conjoined twins are monovular and monoamniotic and therefore identical from both phenotypic and immunologic perspectives.^{12,13}

Studying more than 1200 cases of conjoined twins, Spencer¹² cited this process of “fusion” as the most plausible theory for conjoined twinning. During the third or fourth week of development, the previously separate embryonic discs reunite either dorsally (craniopagus, rachipagus, pygopagus) or ventrally (cephalopagus, thoracopagus, omphalopagus, ischiopagus, parapagus) at sites where surface ectoderm is absent or eventually fuses or opens.¹² These areas include the anlage of the heart (thoracopagus) and diaphragm (omphalopagus), the oropharyngeal (cephalopagus) and cloacal membranes (ischiopagus and parapagus), the neural tube (craniopagus, rachipagus,

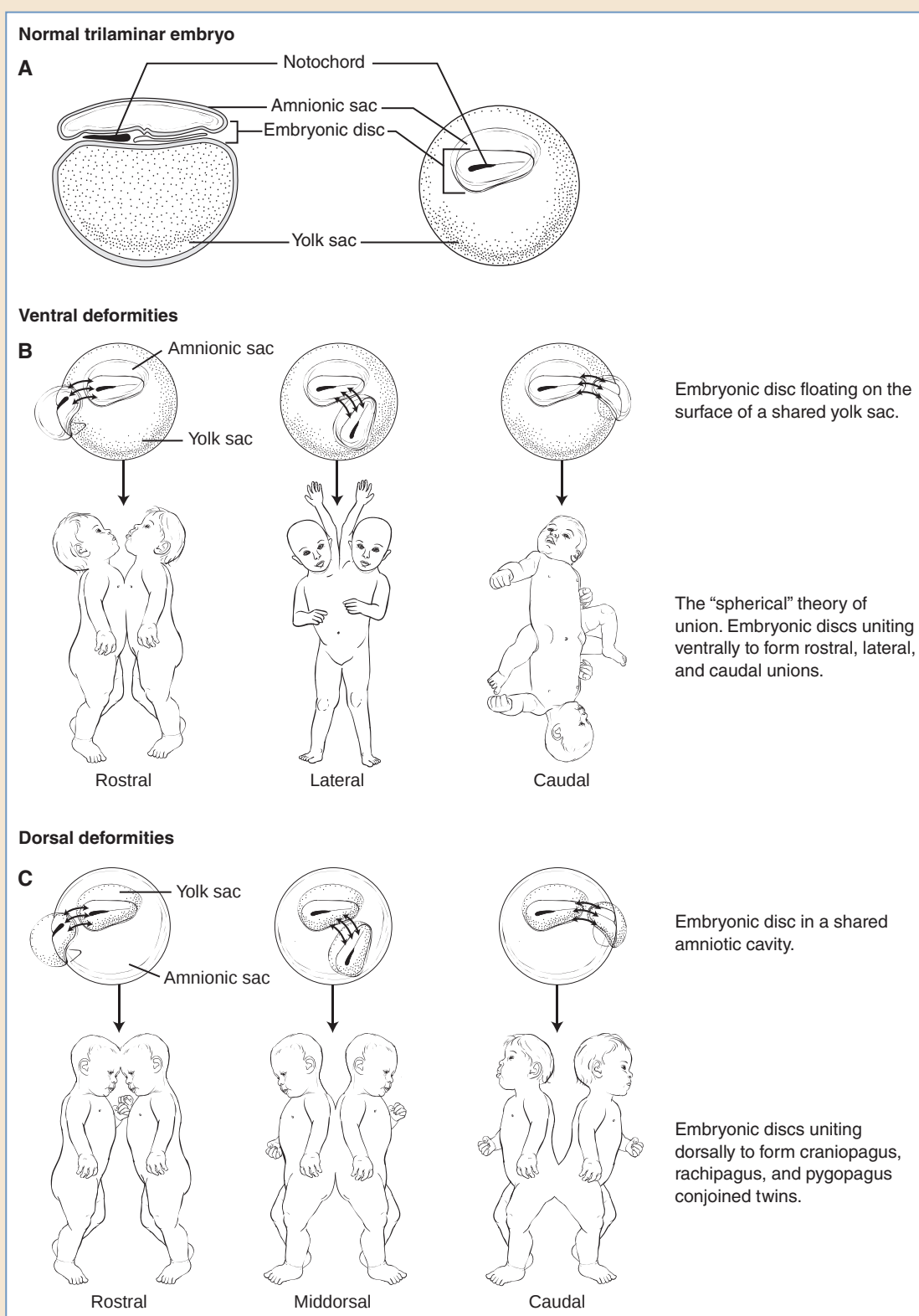


Fig. 14-4 Union of two embryonic discs. **A**, Development of normal individual trilaminar embryo at 3 to 4 weeks. **B**, Embryonic discs reuniting at open areas with subsequent ventral conjoined unions. **C**, Embryonic discs reuniting at open areas with subsequent dorsal conjoined unions.

and pygopagus), and the periphery of the embryonic discs.¹² The union is always homologous (for example, head to head or tail to tail) and can be oriented from rostral to caudal, depending on the relative temporal-spatial relationship of the embryonic discs at the time of fusion (Fig. 14-4).

EPIDEMIOLOGY

According to The International Clearinghouse for Birth Defects Monitoring Systems and the Birth Defects Monitoring Program (BDMP),^{14,15} the estimated mean rate of conjoined twinning is 1 to 1.3/100,000 births, with a significant difference between nonwhites (1.59/100,000) and whites (0.923/100,000), although monozygotic twinning appears to be constant among various ethnic groups. Less than 1% of all monozygotic twins are conjoined.¹⁶ There appears to be no effect of maternal age or parity on conjoined twinning^{15,17}; 40% to 50% of conjoined twins are spontaneously aborted.^{14,15} The majority of conjoined twins are female, with a female/male ratio of 3:1.^{14,15,18,19} Although older data suggest most conjoined twins are born in the spring, more recent studies demonstrate no seasonal trends.^{15,18}

Källén¹⁹ conducted an epidemiologic study of conjoined twins and reported the following information: twins joined in the midportion of the body (thoracopagus, omphalopagus, rachipagus, parapagus) are the most common, with thoracopagus twins representing the majority (30% of all conjoined twins); only 12% of conjoined twins are joined cephalad (craniopagus, cephalopagus); caudal junction (ischiopagus, pygopagus) accounts for 25% of conjoined twins; parasitic twins represent 6%; and malformations include facial clefts, anencephaly, spina bifida, esophageal or gut atresia, gastroschisis, omphalocele, and limb reduction. Data on drug exposures early in pregnancy indicate that thyroid supplementation and the use of prochlorperazine and infertility medications such as clomiphene may be associated with conjoined twinning.^{14,19-21} A recent study by Steinman¹⁶ reported an increased risk of conjoined twinning following the use of oral contraceptives.

NOMENCLATURE

Several classification systems describe the various patterns of conjoined twins. All incorporate the most prominent site of union, followed by the Greek suffix *-pagus*, meaning “joined.” We recognize eight types of conjoined twins, as described by Spencer¹² (Box 14-1 and Fig. 14-5).

Box 14-1 Types of Conjoined Twins

Ventral

Rostral

1. Omphalopagus (umbilicus)
2. Thoracopagus (chest)
3. Cephalopagus (head)

Caudal

4. Ischiopagus (hip)

Lateral

5. Parapagus (side)

Dorsal

6. Craniopagus (helmet)
7. Rachipagus (spine)
8. Pygopagus (rump)

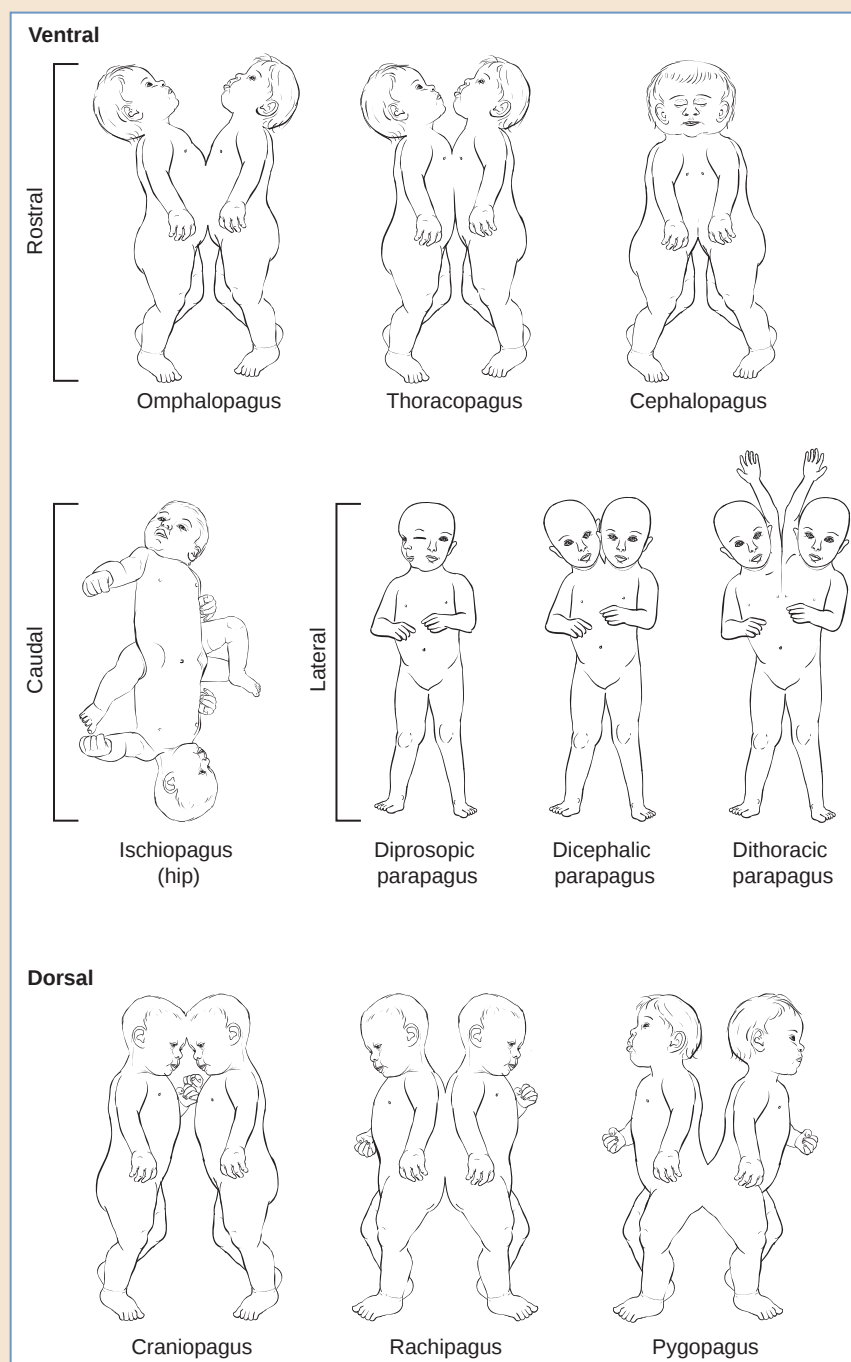


Fig. 14-5 Classification of conjoined twins.

In addition, the prefixes *di-*, *tri-*, and *tetra-* are used to describe the number of shared anatomic structures when they precede *-prospus* (face), *-brachius* (upper extremity), and *-pus* (lower extremity). We find this classification system unambiguous and concise, thus permitting the accurate exchange of important anatomic information when discussing the surgical management of conjoined twins.

Craniopagus

Craniopagus twins represent 2% to 6% of conjoined twin deliveries^{11,22} (Fig. 14-6). Approximately one set of craniopagus twins is born per 2 million live births.²² As with all conjoined twins, craniopagus twins are predominantly female (80%).²² Other intracranial and extracranial abnormalities are common in craniopagus twins.²³

The embryologic fusion of craniopagus twins involves the skull and primitive neural folds.²⁴ Craniopagus twins likely form after the fourth week of development by fusion of two separate embryonic discs at the cranial end of the open neuropore.²³ They can share scalp, calvarium, dural sinuses, and surfaces of the brain; however, the face, foramen magnum, and spine remain separate.²⁵ Varying degrees of interaxis and rotational alignment at the site of union can produce a wide range of anatomic orientations. Facial and skull asymmetry can be present.²³ The exact site of junction and the tissue layers involved also vary. Winston²² reviewed all 79 reported craniopagus twins from 1496 to 1987 and proposed a classification system based on the embryologic origin of the shared tissue (deepest shared structure) (Box 14-2). Craniopagus twins sharing

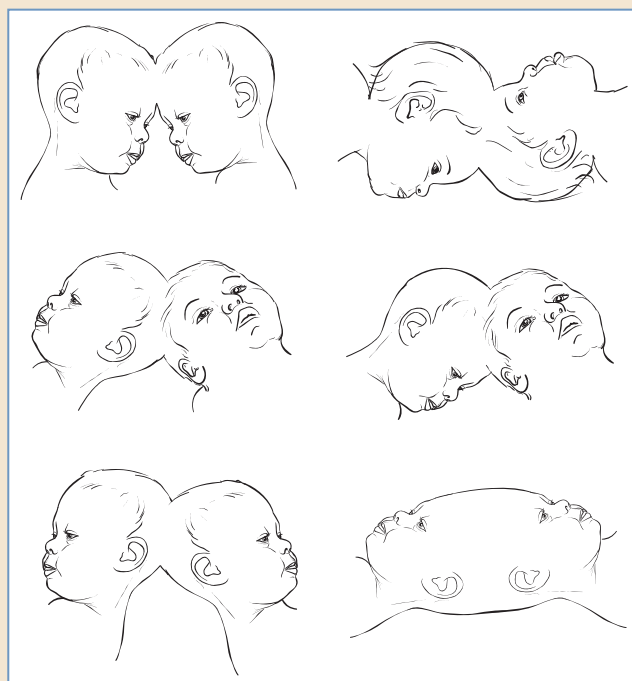


Fig. 14-6 Craniopagus twins.

Box 14-2 Winston Classification of Craniopagus Twins

- Type A: Junctions of scalp, subcutaneous tissue, and possibly bone (least common)
- Type B: Junctions of dura mater (least common)
- Type C: Junctions of leptomeninges (most common)
- Type D: Junctions of contiguous central nervous system (brain)

scalp, subcutaneous tissue, and possibly bone are referred to as having type A junctions. Those who share dura mater are type B. Types A and B are the least common types. Type C craniopagus twins share leptomeninges and are the most common type.²² Type D craniopagus twins have a contiguous central nervous system (that is, brain). Type D junctions occur in 10% to 15% of craniopagus twins.

Craniopagus twins who have contiguous cerebral cortices demonstrate completely independent behavior.²⁶⁻²⁸ Electroencephalographic evidence confirms independent electrical activity, suggesting there are no shared neuronal pathways, even in twins who have continuous central nervous systems.^{22,29,30}

It is important to determine the status of the dural sinuses before attempting surgical separation. A shared dural sinus can be difficult to reconstruct and can greatly affect morbidity and mortality. Blood loss can be massive, resulting in significant neurologic impairment. Attempts at dural sinus reconstruction using cardiopulmonary bypass and hypothermic circulatory arrest have been largely unsuccessful.³¹

The first successful separation of craniopagus twins in which at least one child survived was performed in 1953 by Dr. Herbert Grossman.²⁷ Preoperative studies consisted of electroencephalography, pneumoencephalography, and carotid angiography. Because tissue expanders were not yet developed, long, narrow scalp flaps were “delayed” over a 6-month period to improve their vascularity and venous drainage before rotation. The first attempt at surgical separation occurred when the twins were 14 months old but was aborted intraoperatively because of high blood loss and technical difficulties. The final separation was completed a month later, and the wounds were closed with the previously delayed skin flaps and split-thickness skin grafts. Both children survived the operation, but one died a month later as a result of irreversible neurologic injury and overwhelming infection.

Hoyle³² reported 26 cases of attempted surgical separation of craniopagus twins. The majority of these involved no cerebral connection. The perioperative mortality rate for cases with conjoined cerebri was 61%. Approximately 50% of the twins with no cerebral connection survived separation. Emergent separation was associated with nearly 80% mortality.

Spencer²³ studied 30 attempts to separate craniopagus twins. The majority of these children had large parietal unions with shared venous sinuses. Postoperative complications included neurologic deficits, subdural fluid collections, and hydrocephaly. Half of the twins died, and 17 had neurologic impairment. Only seven children were normal at the time of follow-up.

Cephalopagus

Sometimes confused with craniopagus twins, cephalopagus twins are instead conjoined ventrally in the cephalic portion of the body as a result of a union of the oropharyngeal membrane^{23,25} (Fig. 14-7). The union extends from the head to the umbilicus and includes the brain, foregut, heart, diaphragm, and liver.²⁵ Cephalopagus twins have two faces on opposite sides of the head. One face may be incomplete. A rare form of conjoined twins, cephalopagus twins occur at a frequency of 1 in 3 million births.³³

Omphalopagus and Thoracopagus

Omphalopagus (Fig. 14-8) and thoracopagus (Fig. 14-9) twins can be differentiated by the status of the heart. Both are joined ventrally from the thorax to the umbilicus, but thoracopagus twins always share a heart. Each twin has two arms and two legs and a separate pelvis. Omphalopagus and thoracopagus twins have varying degrees of liver and upper gastrointestinal tract fusion.

In most cases of thoracopagus twins, separation is declined because of the shared heart. In situations in which emergency separation is attempted, the death of at least one twin usually ensues.³⁴⁻³⁶ Elective separation can be performed if the cardiac defect can be reconstructed to allow survival of one twin.

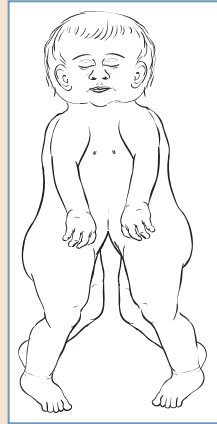


Fig. 14-7 Cephalopagus twins.



Fig. 14-8 Omphalopagus twins with independent hearts.



Fig. 14-9 Thoracopagus twins with a shared heart.

Many of the reported naturally occurring deaths of thoracopagus twins happen very shortly after birth.³⁶ Recent reports describe the successful use of a procedure called *ex utero intrapartum treatment* (EXIT) to ensure adequate uteroplacental gas exchange and hemodynamic stability in a pair of thoracopagus twins during the birth process.^{34,37} In this procedure, the twins are delivered through cesarean section, and the airway is established before clamping the umbilical cord. These results are encouraging and may provide a means of improving perinatal survival in cases of shared cardiac structures in which separation is considered appropriate.

Parapagus

Previously called dicephalus, parapagus twins are united laterally and share a common pelvis (Fig. 14-10). They represent 11% of all conjoined twins.⁶ Their union can extend cephalad and can include the entire abdomen (dithoracic parapagus), the thorax with two separate heads (dicephalic parapagus) (Fig. 14-11), or the head with two separate faces on the same side of the head (diprosopic parapagus).³⁸ Many dicephalic parapagus twins have conjoined hearts, making surgical separation or long-term survival difficult.³⁹ Shared spinal cord innervation will result in significant neurologic deficits if separation is undertaken.³⁴ Many of these twins are offered only supportive care and die shortly after birth.^{34,39}

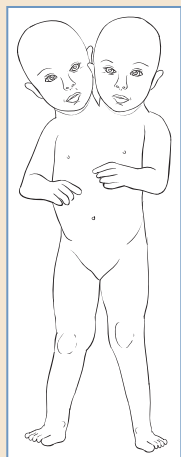


Fig. 14-10 Parapagus (dicephalus) twins.



Fig. 14-11 Dicephalic parapagus twins.

Ischiopagus

Ischiopagus twins present true multidisciplinary challenges when planning separation (Fig. 14-12). United ventrally, they frequently share a liver, gastrointestinal tract, genitourinary system, and pelvic bones, requiring the participation of general surgeons, urologists, and orthopedic and plastic surgeons. The surgeries tend to be lengthy and demand an experienced team of anesthesiologists, surgeons, and nurses. Ischiopagus tripus (three legs) conjoined twins each have a normal leg and a shared malformed third leg, frequently with dual vascular and nerve supply. The soft tissue of the third leg is usually used to cover the soft tissue defect of one of the twins, leaving each twin with one viable leg. Zuker et al⁴⁰ successfully transplanted the entire hindquarter of one dying ischiopagus twin to her sister, thereby leaving her with two “normal” legs. This was the first reported successful lower extremity transplant.

Pygopagus

According to the fusion theory of embryogenesis, pygopagus twins form from the union of two separate embryonic discs in the caudal portion of the open neuropore and can therefore share a buttock, sacrum and coccyx, distal gastrointestinal tract (one anus, two rectums), and part of the genitourinary system^{23,41} (Fig. 14-13). Separation frequently requires preoperative tissue expansion with the subsequent expander capsule being used to reinforce the dural closure if there is a shared spinal cord.⁴¹ Pygopagus twins represent 18% of all conjoined twins.⁴¹



Fig. 14-12 Ischiopagus twins.

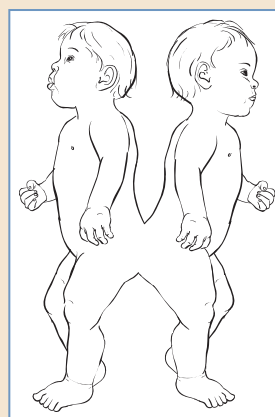


Fig. 14-13 Pygopagus twins.

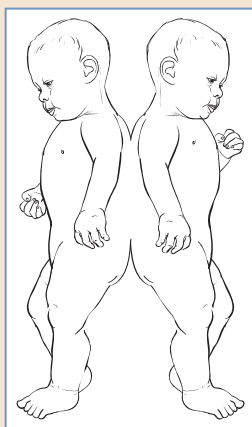


Fig. 14-14 Rachipagus twins.

Rachipagus

This rare type of conjoined twin is united dorsally (Fig. 14-14). In an extensive review of 1200 conjoined twins, Spencer⁴² identified only one typical case and 20 dorsal parasitic twins that were reclassified as rachipagus twins. The one typical intact rachipagus twin case was joined from the occiput to the sacrum, but the bony union consisted only of the thoracolumbar vertebrae.²⁵

ETHICAL CONSIDERATIONS

Moral, ethical, legal, and religious issues frequently complicate the separation of conjoined twins. The parents' and patients' right to privacy, presumed surgical consent, the allocation of organs and genitalia, and the potential sacrifice of one twin for the benefit of the other make the decision to separate some twins difficult. In addition, quality of life and functional outcome with and without separation are important considerations. We have found that including a medical ethicist in the team can be helpful when making such challenging decisions with the family. The separation of conjoined twins almost always attracts a circuslike media atmosphere. Every attempt should be made to respect the parents' and patients' right to privacy.

Several religious and legal considerations are worthy of review. A recent British High Court decision allowed the separation of conjoined twins against the parents' wishes when it was deemed necessary to save the life of one child. The ischiopagus twins involved were Jodie and Mary, born in England in August 2000. Mary had a hypoplastic, poorly functioning heart and was dependent on Jodie for perfusion. The parents were of a strong religious background and "could not begin to accept or contemplate that one of their children should die for the other to survive."⁴³ The English Court of Appeal granted proxy consent to the hospital, and surgery was performed. The weaker twin, Mary, died within 20 hours of the start of surgery. Although "reasonable attempts to separate conjoined twins are supported by both law and ethics,"⁴⁴ every effort should be made to make the decision whether to separate the twins through discussion with the parents.

SURGICAL SEPARATION

Our technical expertise and understanding of conjoined twin physiology continue to improve, and the separation of conjoined twins is no longer experimental. Large medical centers have gath-

ered enough experience that a properly planned and executed operation can usually end with the expected results. Although the decision to separate twins when both are intended to survive is seemingly “easy,” very few if any long-term studies evaluating functional outcome after surgery are available. Many of these children are left with significant neurologic deficits (especially craniopagus twins) and functional limitations as a result of lower extremity deficiencies.

Approximately 50% of conjoined twins die within the first day after birth.³⁴ Twins who require emergent separation have survival rates of 30% to 50%, and those who go on to elective operation have a survival rate of 80% to 90%.^{36,37,45} In a series by O’Neill et al,³⁵ nearly 40% of twins who survived separation died within 10 years from other serious associated congenital anomalies.

Historically, the separation of conjoined twins was thought to be “above human skill.” German cartographer Sebastian Munster⁴ described a set of craniopagus twins separated after the death of one twin at 10 years of age in 1495. The remaining twin died shortly thereafter from the wound she sustained during the separation.

The first attempt at separation of living conjoined twins is said to have occurred in 1689, but documentation of the event is scant.^{32,46} The procedure reportedly involved applying a constricting ligature across a thin band of tissue that connected omphalopagus twins.³² It is believed that the first confirmed successful separation was undertaken in 1902 with 12-year-old omphalopagus twins who shared a small hepatic bridge.⁴⁶ Their separation was prompted by the development of tuberculous peritonitis in one of the twins. Both children survived the operation, but the twin with peritonitis later died of her disease.

Surgical outcome after separation of conjoined twins has improved since the first modern-day separations were attempted in the 1950s.³² Absolute success rates are difficult to determine, because one twin is sometimes allowed to die during surgery. The separation of conjoined twins where both are intended to survive is most successfully performed in ischiopagus twins and least successfully in thoracopagus twins.³²

Emergent separation of conjoined twins carries a higher mortality rate than elective separation preceded by planning and coordination. Preoperative planning and coordination of various surgical teams are central to a successful separation. In this way the extent of various defects can be more adequately appreciated. Stage-setting procedures can be incorporated into the overall treatment plan. To provide additional tissue for coverage, tissue expansion may be used. Intraperitoneal tissue expansion has been described by Zuker et al⁴⁷ to provide not only adequate surface coverage but also adequate abdominal and pelvic support.

IMAGING

Prenatal

Prenatal diagnosis and detailed evaluation helps to prepare the parents and treating team for the birth of conjoined twins. By accurately defining the anatomy and associated malformations, important decisions can be made regarding counseling the parents and planning for the delivery and postnatal course. Useful imaging tools include ultrasonography, echocardiography, CT scanning, and ultrafast fetal MRI.³⁴

Prenatal diagnosis of conjoined twins can be made using ultrasonography during the first trimester. Several sonographic findings are used to aid in the diagnosis of conjoined twins.^{17,48} Ultrasound findings suggestive of conjoined twins include the lack of a separating membrane between the twins, more than three vessels in the umbilical cord, and the inability to separate the fetal bodies.^{17,48} A formal prenatal evaluation using ultrasonography, MRI, and echocardiography should be performed after 18 weeks’ gestation to further define the anatomy and presence of other congenital anomalies.³⁴ Serial sonographic imaging is performed throughout the pregnancy.

Postnatal

CT with three-dimensional bony reconstruction is helpful postnatally for planning surgical separation. CT also provides useful information regarding the complex anatomic relationship of shared organs. The less invasive MRI is preferred to angiography when delineation of the vascular system is needed. As our personal experience increases, we are less dependent on barium studies of the gastrointestinal tract to determine the configuration of the small and large intestine. We and others have noted common and predictable gastrointestinal patterns associated with different types of conjoined twins.⁴⁹

Pregnancy

After a diagnosis of conjoined twins is made, the parents are often counseled regarding the option of terminating the pregnancy. Near-term cesarean section, performed in a tertiary care facility, is recommended for the delivery of conjoined twins.¹⁷ A prepared neonatal ICU with pediatric anesthesia and surgery support is required. Accurate prenatal diagnostic investigations will help determine the need for emergent surgical separation after birth.

PREOPERATIVE PLANNING

The planning process for separation of conjoined twins may be relatively simple or extremely complex. Examples of each will be illustrated in the following cases. Before the surgical separation, a multitude of issues must be resolved. The children must be in optimal condition from a nutritional and developmental standpoint. All ethical decisions regarding separation and the potential for complications must be disclosed to the family. It can be helpful to involve psychologists, social workers, and ethicists in difficult cases in which one of the children may die or be significantly impaired after separation.

Surgical planning should include the various surgical subspecialties, anesthesia, nursing, and the ICU. We include the facility engineers to ensure an adequate power supply, outlets, and overhead lights for two or more teams. We have found that a series of multidisciplinary meetings and an open discussion of each subspecialty's concerns is extremely helpful in anticipating and jointly solving the many concerns that may arise. With complex surgical separations it is also helpful to



Fig. 14-15 Specially designed operating room table for separating craniopagus twins.

have a “run-through.” This is performed in the operating room where positioning of the children and movement of the operating tables can be appropriately planned. We have used a specially designed operating room table when separating craniopagus twins (Fig. 14-15). An electrocautery pad should be placed on either twin at the beginning of surgery to ensure adequate grounding once they are separated.

Anesthesia

Two teams of anesthesiologists are required for any surgical separation. They must be given time and space to provide an adequate environment for the surgery. Major issues such as central lines, intubation, assessment of the common circulation, and provision of adequate volume replacement must be addressed. Anticipation of the logistics of the separation is also essential so that lines do not get entangled or twisted at the time of movement of one child to another site. This is when a run-through is often helpful in anticipating such problems.

The Role of the Plastic and Reconstructive Surgeon

The plastic and reconstructive surgeon will help plan the surgical approach to separation and provide reconstruction of soft tissue defects. This may be relatively simple as with the lesser degrees of omphalopagus twins, or it may be extremely complex as with ischiopagus twins. Soft tissue replacement may involve skin only but also often involves support structures for the abdominal wall, pelvic floor, and even the cranium. In complicated cases it is very helpful to have models to help in the planning process (Fig. 14-16). To provide adequate soft tissue, expansion techniques may be needed. The use of older concepts such as pneumoperitoneum have been replaced with the use of tissue expansion.

As with other planning procedures, planning in reverse is essential—anticipating the potential for problems. Minor wound breakdown should be anticipated and backup plans for coverage prepared. As with other wound-healing problems, provision of adequate nutrition, adequate antimicrobial coverage, and patience to allow for secondary healing will provide the foundations for success.

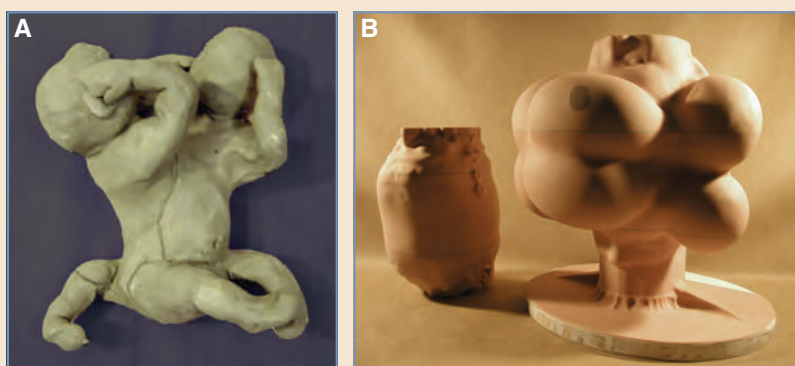


Fig. 14-16 Models can be very helpful in planning the separation of conjoined twins. **A**, Ischiopagus clay model. **B**, Craniopagus twin models before and after tissue expansion.

CASE EXAMPLES

The following case examples illustrate the principles of surgical separation of conjoined twins. Each of these relatively rare procedures must be assessed and planned independently.

Craniopagus Twins

An ultrasonographic examination during late pregnancy showed conjoined craniopagus twins and a third independent fetus. The three babies were delivered successfully through cesarean section in Karachi, Pakistan. The third baby was unaffected. Separation of the conjoined babies was requested, and the infants were transferred to Toronto under the care of a senior neurosurgeon at our hospital. The site of union was at the vertex of each skull with a slight 30-degree rotation (Fig. 14-17, *A*). There was a shared segment of cerebral cortex and a shared sagittal sinus. The cardiac status of one baby was excellent, whereas it was quite poor in the second. This became very important regarding the timing of separation surgery. The gastrointestinal systems were normal in each child. One baby had two normal kidneys, whereas in the other baby one kidney was absent and the other kidney was severely hydronephrotic and nonfunctional, indicating the entire renal filtration mechanism was limited to the baby with two normal kidneys. Separation would leave the other baby with no renal function.

After considerable discussion involving religious leaders, ethicists, and psychologists, it was decided to transplant one kidney from the healthy baby to the second. This became a significant ethical problem in that, by Ontario law, transplantation from a live donor is not allowed until the age of 18. However, the conjoined twins were legally considered one entity, and thus transplantation from one part of the single entity to another would be allowed. The kidney transplantation was performed successfully 6 months before the separation.

Cerebral circulation was investigated, and attempts were made to partially close the shared venous drainage systems so that each baby would have appropriate independent venous drainage. This was undertaken through embolization techniques, which had not been done previously.⁵⁰ The conjoined cranial surface was broad, indicating a significant need for scalp and calvarial reconstruction. Most of the bony structures could be reconstructed with split calvarial bone grafts, but we would need additional tissue expansion to provide sufficient scalp coverage. Two tissue expanders were placed on either side of the conjoined cranium (Fig. 14-17, *B*).

The second twin's cardiac status deteriorated, necessitating urgent separation before tissue expansion was complete. After the scalp flaps were elevated, a strip of conjoined cranium was removed circumferentially. This provided access to the brain for the neurosurgeons and was used in the calvarial reconstruction. Despite prior embolization efforts, the separation was difficult and blood loss was high. The cerebral structures were separated and the large dural defects reconstructed with dural grafts. The split calvarial grafts were used for skull reconstruction. The scalp flaps were repositioned and full coverage was achieved (Fig. 14-17, *C*). The babies were transferred to the PICU and extubated 48 hours after surgery. There were some areas of scalp wound breakdown that necessitated wound care and eventual skin grafting. One child never fully regained consciousness and died 2 weeks after surgery. The cranium of the surviving twin coalesced nicely, and the scalp flaps survived and produced luxuriant hair growth. There were some areas of alopecia at the site of the skin grafts and at the sites of scalp flap union. Over the course of the next month, the baby was eating normally and functioning at the appropriate developmental level (Fig. 14-17, *D*). She returned to her native Pakistan with her parents and the other unaffected triplet.



Female Omphalopagus Twins

A Canadian family living in Mexico was found to be expecting conjoined twins during routine ultrasonographic examination. The mother was transferred to our hospital, where the children were delivered at 38 weeks' gestation through cesarean section. Both girls were healthy and classified as omphalopagus twins (Fig. 14-18, A). Ultrasound evaluation identified a 6 cm conjoined union consisting of a segment of shared liver. Evaluation of the biliary systems found normal biliary tracts going from each baby's main liver to the same baby's small bowel. Separation was deemed to be fairly simple, consisting of skin and abdominal wall coverage and division of the shared liver. It should be noted that on occasion the biliary systems can be crossed. In such cases, repositioning the biliary drainage system is necessary for adequate hepatic function.

A team of general surgeons, plastic surgeons, anesthesiologists, intensivists, and nurses was assembled. The general surgeons led the team, because they were responsible for separating the liver. The liver was separated, and confirmation of biliary drainage for each baby was ensured. On separation one baby was placed on a second operating table for reconstruction of the abdominal walls. The abdominal walls were closed primarily in layers without difficulty. Umbilical

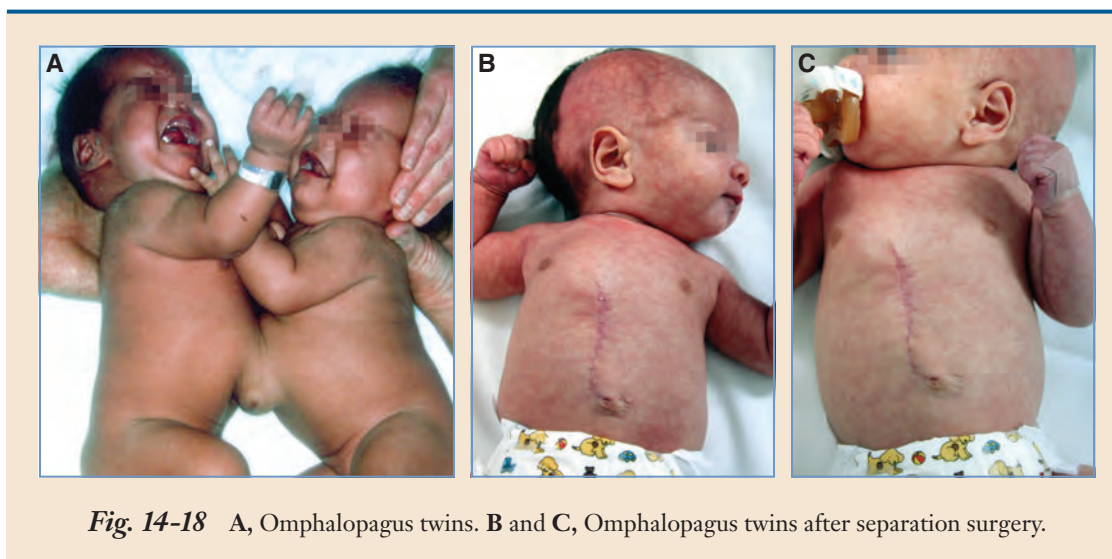


Fig. 14-18 A, Omphalopagus twins. B and C, Omphalopagus twins after separation surgery.

reconstruction was also performed (Fig. 14-18, B and C). The children were transferred to the PICU, extubated on day 1, and transferred to the pediatric surgery ward on day 3. They remained in the hospital for an additional 10 days to ensure adequate gastrointestinal function and appropriate wound healing. The children were discharged home and have developed normally, with no significant abdominal wall deformity.

Male Omphalopagus Twins

A woman in rural Zimbabwe was found to be carrying omphalopagus conjoined twins on routine ultrasound examination. The babies were delivered without complication at 37 weeks' gestation and found to be healthy. They had a large area of union from the xiphoid process to just below the umbilicus. They were also noted to have unilateral complete cleft lip and palate. After nutritional stabilization they were transferred to our hospital (Fig. 14-19, A).

Detailed investigations demonstrated a shared liver with separate biliary systems, intestinal tracts, and genitourinary systems. Despite a significant chest wall deformity, the hearts were separate.

Because of the family's socioeconomic needs, a variety of volunteer organizations helped with the transportation, housing, and hospital costs. All medical fees were waived. The children underwent nasogastric feeding to improve their nutritional status for 3 months before separation. Plans for separation were made, and a variety of team meetings were held.

As in the case shown in Fig. 14-17, the liver would be separated and confirmation of appropriate biliary drainage would be ensured. The children's union was very broad, involving the lower chest wall and abdomen. At 5 months of age, a large tissue expander was inserted over the conjoined section of the thoracic cavity. The expansion progressed satisfactorily until week 10, when the expander became infected and eroded through the skin. The expander was unsalvageable and was removed. Because all of our expansion was lost, we would need to use artificial fascia for the fascial defects and regional skin flaps for the skin defect. With the help of three-dimensional imaging, our surgical plan included an incision that was somewhat offset to provide for direct soft tissue coverage for one baby. For the second baby we would require a regional flap to cover the essential areas of the lower chest and upper abdomen.



Fig. 14-19 A, Omphalopagus twins. B, Closure of twin A. C, Closure of twin B. D, The boys after recovery from separation surgery.

Surgical induction was undertaken without incident, and our surgical incisions were outlined. It is helpful to staple along the planned incision lines, particularly for the back, because ink marks tend to be obscured during the lengthy separation process. There was fibrous and cartilaginous adherence of the thoracic cages, and care was taken to separate these without entering the pleural cavities. The two hearts were normal and unaffected. On entering the abdominal cavity, we identified a large segment of conjoined liver. This was separated, the posterior component was incised and separated, and one baby was moved onto another operating table for closure.

The incision on the baby with more fascia and skin was closed with an inferior fasciocutaneous flap (Fig. 14-19, B). Closure was effected transversely just below the costal margin. This was a secure fascial closure as well as an adequate skin closure. Relatively large dog-ears were left laterally to be addressed later, at the time of cleft lip and palate closure. The twin with the lesser amount of fascia and skin (Fig. 14-19, C) posed a more difficult reconstructive challenge. We were able to mobilize the fascia over the lower abdomen to achieve a secure abdominal wall closure. The fascial defect over the liver was closed with SurgAssist and covered with a large transposition flap from the lower abdomen.

The children were then returned to the PICU and extubated 48 hours after surgery. They were transferred to the pediatric surgery ward on postoperative day 4. They remained hospitalized for an additional 2 weeks. Normal feeding was undertaken within 1 week, and wound healing progressed satisfactorily. Distally, a 2 cm area of necrosis was present on the transposition flap, which healed with adequate nutrition, wound care, and patience.

Both babies recovered in a home setting for an additional month. They were returned to the operating room at 8 months of age for cleft lip and palate repair. Six weeks later they were discharged home (Fig. 14-19, *D*).

Ischiopagus Twins

A late-term ultrasound examination performed on a patient in Trinidad revealed a set of conjoined twins united at the pelvis. These ischiopagus twins were born without incident by cesarean section. They were healthy boys with two legs each but shared a single penis. The site of union was just caudal to the umbilicus. The classification for this set of twins was tetrabrachius, tetrapus, ischiopagus conjoined twins (Fig. 14-20, *A*).

A multitude of diagnostic tests were performed, including ultrasonography, CT scanning, standard radiographs, and barium studies of the gastrointestinal tract. The gastrointestinal tracts were separate up to the foramen cecum, at which point the bowel was united. Genitourinary studies demonstrated that one kidney drained through a ureter into the other baby's bladder. There was a single large bladder with a central partially formed septum. Each baby had intraabdominal undescended testes. The penis was shared but had four corpora. This was important, because it allowed separation of the penis into two structures. In certain sets of ischiopagus conjoined twins, the shared penis has only two corpora and cannot be divided. In one other case of ischiopagus twins in which the penis had only two corpora, we elected to create a neovagina using sigmoid colon to gender-convert one of the boys to a girl. We have had one case in which the gender-converted boy to girl opted for remasculinization of her genitalia when she became a teenager.

In this set of ischiopagus twins we elected to place five subcutaneous tissue expanders and two intraperitoneal tissue expanders to provide skin and soft tissue coverage for the abdomen and pelvis.⁴⁷ The intraabdominal expanders were placed through a posterior flank incision. The babies also required a colostomy, because there was no anal orifice. The tissue expander was inserted at the Port of Spain General Hospital by the senior surgeon, and inflation of the expanders was performed by the local plastic surgeon. Expansion progressed satisfactorily. After 6 weeks, feeding became difficult because of the enlargement of the intraabdominal expanders. Frequent small feedings were necessary to maintain nutritional support. Three months after tissue expander insertion, the children were transported to our hospital for further assessment, diagnostic procedures, and definitive separation surgery (Fig. 14-20, *B*). As is always the case with ischiopagus twins, CT revealed that the infants shared a single pelvic ring. Both the upper and lower extremities were normal. Detailed planning sessions were carried out so that each service knew the problems and concerns of the others. A run-through procedure was performed.

After positioning the patients and induction of anesthesia, our markings were made and traced with a stapler to ensure that they were not rubbed off during the first part of the operation. Ventilation of the lungs was difficult because of the large intraabdominal tissue expanders; these were removed at the start of surgery. The large bowel was separated into two segments. The urologic structures were separated accordingly, and the aberrant ureter was reinserted into the appropriate bladder. The corpora were separated and each child given a reconstructed penis. The orthopedic surgeons then performed pelvic osteotomies to form two separate pelvic rings. Each pelvis had two limbs from the appropriate baby. The final incisions were made on the back, and the twins were separated and placed on separate operating tables.

The reconstructive component was quite difficult and complex, because it required pelvic support, abdominal support, and skin coverage. Our intraabdominal expanders were effective in providing expanded tissue for pelvic support and abdominal support. Of particular importance was the capsule that had formed from the intraabdominal expanders. This was effectively used for abdominal support and complete closure in one of the children. The other child required a Marlex patch to complete closure. The skin flaps were then positioned and closed.

The large incisions from the chest expanders were somewhat dusky at the tip. The babies were then transferred to the PICU, where they remained intubated for 48 hours. Seventy-two hours later they were transferred to the pediatric surgery ward. The skin flaps had crossed the midline, and concern about their vascularity was noted. Each child eventually required a small, split-thickness skin graft on the chest wall once the flaps demarcated and were debrided with appropriate wound care. The small open areas healed uneventfully, and a favorable outcome was achieved (Fig. 14-20, *C* and *D*). One concern that did become a problem later in life for the boys was the positioning of the penis and scrotum. In both babies it was necessary to make this position quite inferior and caudal. However, both boys had effective normal micturition, and limb and pelvic growth seem to have been quite normal (Fig. 14-20, *E*).



Fig. 14-20 A, Tetrabrachius, tetrapus, ischiopagus conjoined twins shortly after birth. B, The twins are shown at approximately 3 months, after inflation of intraoperative and subcutaneous expanders. C and D, The twins are seen postoperatively. E, Late result approximately 6 months after surgery.

Composite Tissue Transplant in Ischiopagus Twins

An ultrasonographic evaluation at 24 weeks' gestation revealed ischiopagus conjoined twins in an otherwise healthy 38-year-old woman. The babies were delivered by cesarean section at 36 weeks' gestation and then transferred to The Hospital for Sick Children in Toronto for care. They appeared to be healthy sisters, with union from the lower chest to the suprapubic region. Each had two normal upper extremities but only one normal lower extremity. There was a single pelvic ring with an additional third lower extremity, which was small, hypoplastic, deformed, and exhibiting minimal function. Thus these twins would be classified as tetrabrachius, tripus, ischiopagus conjoined twins (Fig. 14-21, *A*).

Further cardiac investigations confirmed a severe anomaly in twin A that consisted of a double-outlet right ventricle, a very restrictive ventricular septal defect and atrial septal defect, and pulmonary hypertension. CT scans and MRIs were carried out and demonstrated a shared liver with a separate biliary drainage system for each baby. There was a large urogenital sinus with one functioning kidney in twin A and two functioning kidneys in twin B. However, the right kidney in twin B demonstrated ureteropelvic junction obstruction with a greatly dilated renal pelvis. As has been noted in previous ischiopagus cases, the functioning kidney in twin A drained into the bladder of twin B.

The gastrointestinal tracts were separate until the distal ileum, as is commonly the pattern in ischiopagus twins. A reconstructed three-dimensional CT scan of the pelvis clearly demonstrated the pelvic ring as well as three hip joints and one symphysis pubis.

Elective tissue expansion was planned to start at 6 months of age. However, at 3 months of age, twin A developed acute cardiac decompensation. Several cardiologists and cardiac surgeons were consulted and felt that twin A would succumb to her cardiac anomaly within the next few days, and salvage of any kind was impossible. The cardiac anomaly in twin A was lethal and inoperable and necessitated urgent separation if twin B were to survive. After discussions with the children's family, ethicists, religious leaders, social workers, and team members, the decision was made to proceed with the separation, anticipating the demise of twin A either intraoperatively or shortly thereafter. Plans were made to transplant the normal leg of twin A onto the surviving twin B (Fig. 14-21, *B*).

The procedure began with the anterior abdominal wall incision. The hepatobiliary systems were separated, as were the gastrointestinal and urinary tracts. Twin B retained the gastrointestinal tract in continuity to the anus. We then identified the sciatic nerve coming from twin B to the third shared deformed limb. The nerve was transected and coapted to the sciatic nerve going to the transplanted limb from twin A. We preserved a portion of the myocutaneous tissue on the third shared limb to provide for active hip flexion and knee extension. The anterior thigh mass would be piggybacked on to the anterior thigh of the transplanted limb to allow hip flexion and knee extension. The femoral and iliac vessels, as well as the femoral nerve to this thigh mass coming from twin B, were left in continuity, providing an independently perfused and independently innervated myocutaneous flap of anterior thigh mass.

The pelvic osteotomies were done in such a way as to preserve the symphysis pubis between the two healthy hip joints and to create a single pelvic ring, with elimination of the hip joint from the third shared limb. The separation was complete and the transplanted limb was rotated posteriorly relative to twin B to create a single pelvic ring with two hip joints on either side of the symphysis pubis. The transplanted limb was then revascularized, with the aorta and inferior vena cava from twin A being anastomosed in an end-to-side fashion to the aorta and inferior vena

cava of twin B just below the level of the renal vessels. The stump of the sciatic nerve from twin B, which had previously innervated the third shared limb, was coapted to the sciatic nerve of the transplanted limb that had previously been innervated by twin A (Fig. 14-21, C and D).

After revascularization and reinnervation of the transplanted limb, the myocutaneous flap was sutured to the quadriceps tendon of the transplanted limb. The abdominal and chest flaps were trimmed and adequate fascial support was provided to the abdomen and pelvis of twin B.

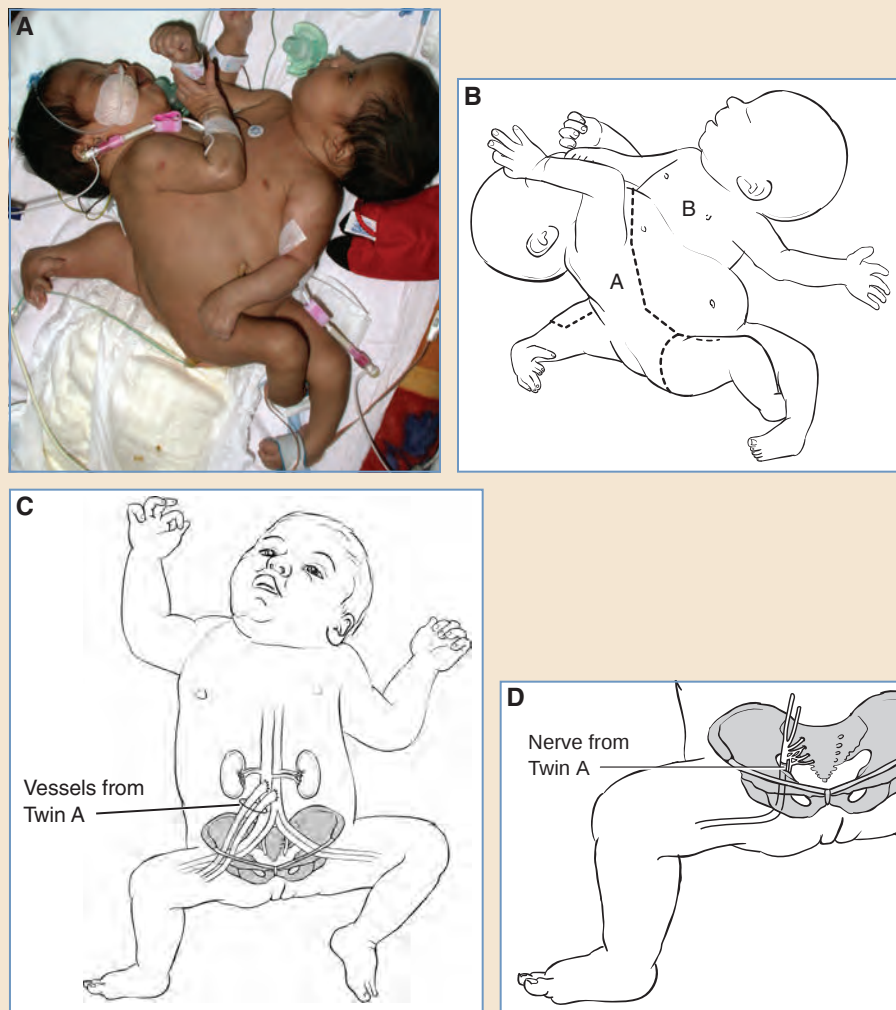


Fig. 14-21 A, Tetrabrachius, tripus, ischiopagus twins. B, Planning for limb transplantation. C, Revascularization and reinnervation of transplanted lower extremity. D, Surviving twin after revision surgery.

Continued

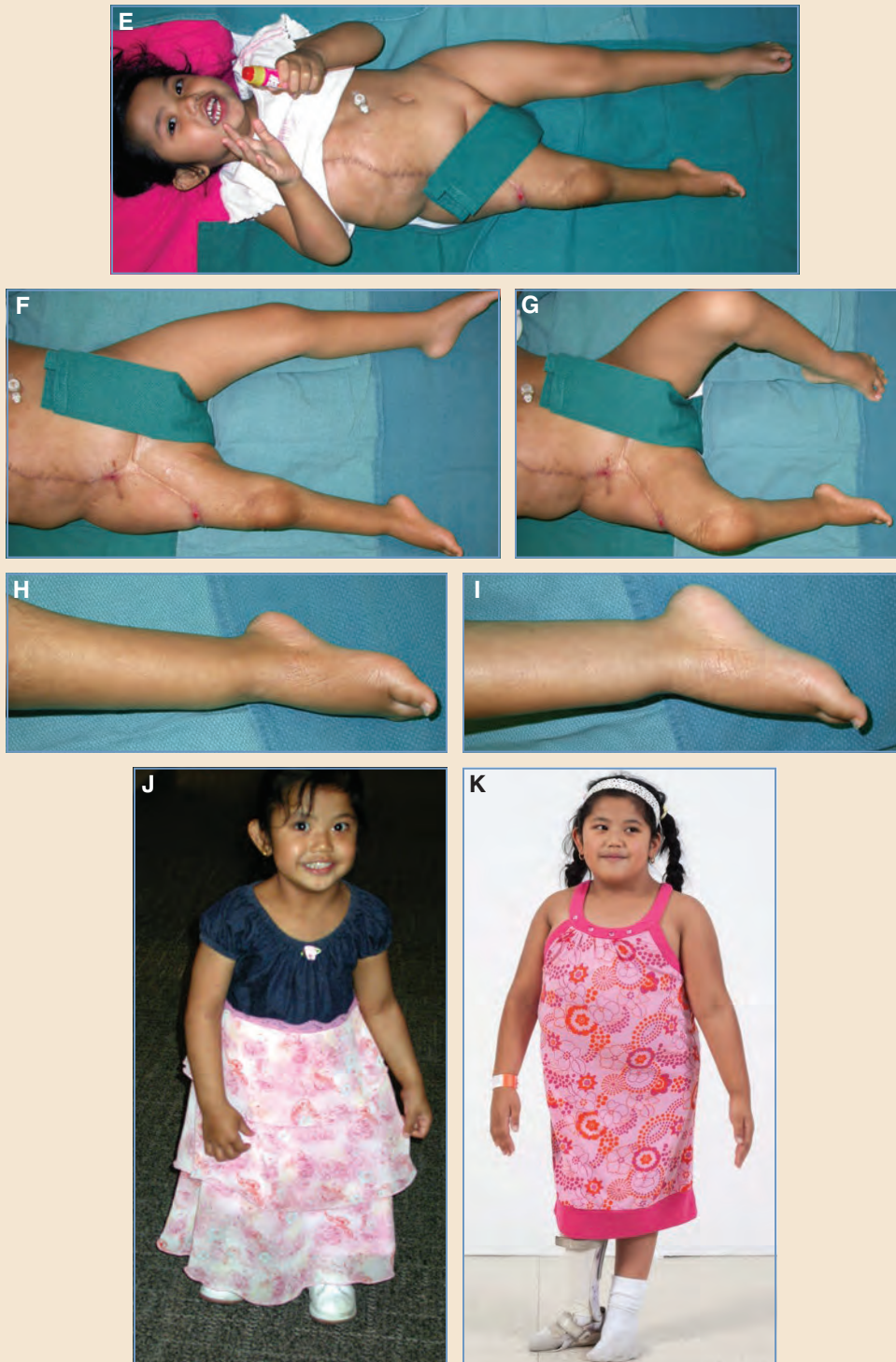


Fig. 14-21, cont'd E, Twin B recovered full sensation in the transplanted limb and had excellent movement. F, Knee extension. G, Hip flexion. H, Plantar flexion. I, Toe movement. J, Limb function has been completely integrated into her normal cerebral function. K, Patient at 6 years of age.

Twin A died shortly after the separation. Twin B was transferred to the PICU, where she remained before being transferred to the pediatric surgery ward. There were no wound-healing problems, and she was discharged home 3 weeks after the separation. She required some minor revision surgery at 33 months of age to reposition the abducted transplanted limb by a femoral osteotomy and to tighten the tendinous insertion of the myocutaneous flap. Simultaneously, the abdominal scar was revised (Fig. 14-21, *E*).

The limb has continued to grow in proportion with the child. She has recovered full sensation to the transplanted limb and has regained excellent movement (Fig. 14-21, *F-I*). The transplanted limb appears to be completely integrated into her normal activities with appropriate voluntary movement on command and spontaneous activity with ambulation (Fig. 14-21, *J* and *K*). It is interesting that the Tocci brothers referred to earlier in this chapter were not able to achieve this, because their cortical-neural connections were separate and thus not integrated.

• • •

The original kidney transplant performed by plastic surgeon and Nobel laureate Joseph Murray used the immunocompatibility of identical twins for its success. Similarly, in conjoined twins, tissue can be transplanted from one twin to another, thus affording numerous transplant opportunities. In this section we have seen the role of kidney transplantation in conjoined twins and split-thickness skin grafts crossed between one twin and the other. In the last case, an entire lower extremity was transplanted from one twin, who was destined to succumb to her cardiac anomaly, to her surviving sister, who would otherwise have had only one leg.

CONCLUSION

These case examples illustrate several principles in conjoined twin separation. First, one should have as much information as possible before undertaking separation surgery. This not only involves detailed evaluations but also may include examinations with the patient anesthetized.

In these complex procedures it can be anticipated that wound-healing problems will arise. Adequate nutritional support is essential, as is intensive care monitoring and support. In one of our ischiopagus twin separations (not reported here), a large area of exposed abdomen was present in one baby. Without intensive care support, this baby surely would not have survived. Patience is extremely important in these cases. Time and adequate nutrition will provide the essentials for wound healing. We have learned not to rush into reconstructive procedures for secondary wound issues but rather to allow granulation tissue to form and then proceed with simpler procedures, such as skin grafting.

We have also learned that even expanded skin flaps do not do well if they cross the midline in conjoined twins. The vascular connections do not cross the midline, and thus one cannot expect large flaps to survive if they are based on the midline. Care in planning and in insertion of tissue expanders is essential.

We have seen that sex change procedures in conjoined twins are not as effective as one might expect. We opted to carry out a sex change procedure in one child, which probably would have been better approached with a penile reconstruction at a later date.

The value and importance of multidisciplinary planning and the involvement of all team members are crucial in conjoined twin separation surgery. This includes team members from anesthesiology, intensive care, and the various subsurgical specialties involved as well as psychologists, ethicists, and particularly social workers. Support for the family is imperative during these complex and high-risk procedures.

The separation of conjoined twins should not be undertaken lightly by any institution. Expert support in all areas must be present, and a coordinated team effort is essential. The risks are high, but the results can be extremely gratifying for the professionals involved and, of course, for the parents, families, and children involved.

KEY POINTS

- It is essential to gather as much information as possible before separation surgery is undertaken.
- The surgical team should be patient and anticipate secondary wound healing.
- The twins' nutritional status needs to be maximized before separation.
- PICU support is essential.
- The site of twin union must not be crossed in flap design and reconstruction.
- Sex alteration should be avoided.
- This is a complex procedure requiring a full team of experts.
- Surgical planning is paramount, and a surgical rehearsal can be valuable in multidisciplinary surgeries.

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15

Soft Tissue Trauma

Andreas M. Lamelas • Peter J. Taub



Young children sustain a disproportionate number of soft tissue injuries to the head and neck region compared with the incidence in adults, largely because of children's curiosity, lack of caution, and still-developing vestibular sense (balance). The mechanisms of injury typically seen in children include falls, sporting activities, motor vehicle collisions, interpersonal violence, and abuse, with the frequency of each cause differing between age groups. In 2010, head, face, and neck injuries to children under the age of 15 accounted for 3.6% and 1.8% of all emergency department visits in males and females, respectively.¹

Because of the rapid growth of their brains, a child has a relatively large head compared with the rest of his or her body, making this area very susceptible to injury. The introduction of several governmental safety regulations, including mandatory airbags, the use of seat belts, child seats, helmets, and mouth guards, has directly led to a decrease in head and neck injuries, but even with such safeguards in place, significant soft tissue injuries occur and present a real challenge to the plastic surgeon. These challenges are often compounded by the potential adverse effects the injury may have on the future growth of the child, both physically and mentally. The face represents the most important presenting part of the body and plays a major role in psychological development. An unfavorable outcome may have drastic effects on the child's interpersonal relations and self-esteem. As a result, counseling of both the patient and the parents is often as important as the surgical treatment of the injury. To successfully manage pediatric soft tissue and maxillofacial injuries, the clinician must have a firm grasp of the physiologic and psychological principles associated with facial injuries.

The goals of management of facial trauma in all age groups are to restore and preserve function while achieving optimal cosmetic results and minimizing scar formation. The pediatric

population requires modified therapies to accommodate their unique differences, including anatomy, rapid healing, fragile psychology, and the potential for future deformity in relation to altered facial growth. Although usually minor in nature, soft tissue injuries are the most common facial injuries in children, encompassing a spectrum that includes bruising, abrasions, lacerations, avulsions, and burns. Fortunately, most of these injuries can be repaired in the emergency department or office with the use of a local anesthetic, but general anesthesia should not be avoided if it will facilitate a better end result.

PATIENT ASSESSMENT AND MANAGEMENT

The evaluation of all patients with head and neck injury should begin with the ABCs of trauma to confirm that the patient has an adequate airway, is breathing appropriately, and is maintaining systemic circulation. Emergent airway stabilization is rarely required with isolated soft tissue injuries, but the need for tracheostomy in these patients with no concomitant skeletal injury is associated with high mortality rates. Craniofacial injuries are identified after the patient is stabilized, and the patient is assessed for intracranial, ophthalmologic, and cervical spine injuries.² Children are often frightened of medical personnel, and the initial contact should focus on gaining trust, reassurance, and achieving mutual cooperation.³

A detailed history should be obtained to determine the nature of the soft tissue injury and the likelihood that there are underlying fractures. If an embedded foreign body or associated maxillofacial fracture is suspected, radiographic studies are indicated. Although plain films may be helpful, such as an orthopantomogram or oblique views of the mandible, CT scanning is the mode of choice to evaluate not only the osseous structures but also the soft tissue. Severe maxillofacial injuries warrant a CT scan of the brain, which allows the team to obtain maxillofacial cuts for clear definition of skeletal injuries.

Soft tissue injuries are classified by the cause, location, size, depth, extent of soft tissue loss, and any important structures involved. Photographs should be taken with the parent's consent to document the injuries. Pretreatment and posttreatment photographs can be very important for addressing insurance, child abuse, and medicolegal issues. If child abuse is strongly suspected, the appropriate authorities must be notified and clinical findings documented in the patient record.

Patient cooperation with a pediatric injury may be difficult. It is essential to have the child calm and motionless so necessary treatment can be provided. Some children do well with gentle reassurance and parental support, but the use of sedation may be necessary in certain circumstances. Infants usually require the presence of a parent as a soothing influence for evaluation. Older children may be calmed with distraction, such as a video on a handheld tablet. Adolescents may benefit from asking the parent to leave the treatment area. Many institutions employ child-life specialists to assist with the care of pediatric patients. Before any intervention, the patient and parent should be informed of the proposed plan, and realistic expectations should be established to allay surprise or dismay after the repair. This includes education on postoperative care, the healing process, and eventual scarring.

Analgesia may be obtained with one or more medications before the repair begins. If necessary, sedation may be safely achieved with agents such as ketamine or propofol. The use of such agents requires close monitoring of the patient's vital signs and avoidance of unintended general anesthesia. If sedation is required, the timing of the patient's last meal should be noted to help prevent aspiration. Most soft tissue injuries in children can be managed in the emergency department, but in certain circumstances, such as the treatment of large wounds or injuries that have disrupted delicate structures, general anesthesia in the operating room may be more appropriate.

For all major injuries, local anesthetic should be administered before wound cleansing and exploration. To minimize the discomfort of an injection, a local anesthetic should be administered

through the margins of the wound rather than into the surrounding skin, and topical anesthetics such as lidocaine/prilocaine (EMLA) are available to further decrease the pain of injection. Regional nerve blocks are very useful in larger injuries, especially those involving the forehead, cheeks, lips, and chin. These blocks minimize the tissue damage and anatomic distortion created from direct injection into the wound. The forehead can be blocked by direct injection of the supratrochlear and supraorbital nerves bilaterally, located just above the superior orbital rim, 1.7 and 2.7 cm from the midline, respectively. The upper lip, lateral nasal wall, and cheek are blocked with injection of the infraorbital nerve, located 4 to 7 mm below the infraorbital rim in line with the medial limbus. The lower lip and chin are blocked with bilateral injection of the mental nerve, located below the apex of the second bicuspid.⁴

After appropriate anesthesia is administered, a detailed inspection of the wound should be performed to identify all structures involved and the presence of any foreign material. Any foreign material must be meticulously removed to prevent future cosmetic deformity, such as tattooing. Devitalized areas should be debrided judiciously to maximize preservation of available tissue. The rich blood flow of the face allows a significant amount of injured tissue to survive, even with a seemingly small vascular bridge. Copious irrigation allows most wounds to be closed immediately, with low rates of infection. In patients with significant tissue loss or gross contamination, a delayed repair may be indicated.

The planning and execution of proper skin closure in pediatric patients is imperative to improve the final outcome. This involves appropriate debridement and irrigation, accurate repositioning and alignment of affected tissues, and the use of the correct closure material. Injuries that extend across the margins of the lips, nostrils, eyelids, or external ears may benefit from tattooing before manipulation to facilitate exact anatomic alignment during closure. In general, deeper tissues, including muscle, fascia, and subcutaneous fat, are closed with absorbable sutures. Closing deeper tissues decreases tension on the skin and minimizes scar formation. Skin closure should be performed with the knowledge that suture removal can be an equally traumatic event for the patient. Many options are available, and the decision is left up to the surgeon. Options include the use of fast-absorbing catgut suture on the skin, placement of subcuticular sutures with overlying tissue adhesives (DermaBond) or Steri-Strips, and recently, the use of pull-through monofilament sutures followed by tissue adhesive application after suture removal. The use of staples is an effective method for wound closure but should be reserved for injuries to the scalp.

TYPES OF INJURY

Hematomas

Hematomas are a sequela of a blunt trauma that has caused rupture of dermal or subdermal blood vessels and can often indicate a more significant underlying bone injury. Small hematomas often resolve with warm compresses and observation; however, large hematomas may require aspiration with a fine needle 7 to 10 days after injury once liquefaction occurs.⁵ Hematomas near the septal and conchal cartilage require specific mention; these will be discussed in their respective sections. Delayed hematomas may not be easily aspirated and if large may need operative evacuation.

Abrasions

Mechanical abrasions represent partial-thickness skin injury and vary in thickness. *Superficial abrasions* localized just to the epidermis should be thoroughly cleaned and covered with nonadhering dressings, such as antibiotic ointment or Xeroform gauze. To adequately cleanse the wound, local anesthesia may be required to reduce pain. Superficial abrasions will heal without a permanent scar with proper cleansing and local wound care. *Partial-thickness abrasions* extend into the dermis.

Conservative management for partial-thickness injuries is paramount and includes judicious debridement of devitalized tissue and scrubbing with antimicrobial soap to remove debris.⁶ Most partial-thickness abrasions will heal with minor scarring. *Full-thickness abrasions* extend through the dermis and into the subcutaneous tissues. These injuries are a result of a significant blunt trauma and should be an indicator that other serious injuries may be present. After proper wound debridement and copious irrigation, primary closure is performed when possible. Although local advancement flaps are often needed to properly achieve wound coverage, the wound should be given time to demarcate in case additional debridement is needed. In all cases it is very important to remove embedded material to prevent permanent tattooing. For full-thickness injuries, antibiotic coverage is strongly recommended.

Lacerations

Facial lacerations can vary significantly, depending on location, size, depth, and associated injuries. At first glance the laceration may seem small, but such injuries often extend deeper and can affect both sensory and motor nerves, the parotid ducts, lacrimal ducts, and other critical facial structures. All lacerations must be thoroughly irrigated, inspected, and properly debrided, usually requiring a combination of anesthetic techniques, depending on the age and affect of the child. Injuries that extend across margins, such as the lips, nostrils, eyelids, or external ears, require particular attention, because even small defects in alignment will be readily noticeable. Periorbital injuries should prompt a thorough eye examination and an ophthalmology consultation to rule out ocular injury.⁶

In general, facial lacerations should be debrided minimally to maximize tissue preservation and allow primary closure. Superficial lacerations require closure of just the dermis and epidermis. A well-approximated deep dermal layer with fine absorbable sutures is essential to decrease skin tension and prevent scar widening, although one study found that superficial, nongaping (less than 10 mm) linear lacerations can be closed in one layer with similar cosmetic results.⁷ The surgeon has many options for skin closure, including absorbable and nonabsorbable sutures or tissue adhesives. Traditional teaching recommended the use of nonabsorbable sutures, because they are minimally reactive and maintain good tensile strength. However, their main disadvantage is that they need to be removed, which can be very difficult and even damaging in a young or very anxious patient. Recent studies assessing both absorbable sutures and tissue adhesives (DermaBond) have found that they are equally effective and provide the same cosmetic results.^{8,9} For deeper lacerations, a layered closure is important to reapproximate functional muscle or fascial planes. Muscle, fascia, and subcutaneous sutures should be a fine absorbable material, such as Vicryl or PDS. Mucosal defects are usually closed with chromic gut, but some surgeons recommend Vicryl, because it is less irritating to the patient. Two of the most common complications of a primary facial closure are infection and hematoma. For this reason, meticulous attention to hemostasis and copious irrigation are crucial before final epidermal closure.

Postoperative care of facial lacerations is often debated and is ultimately up to the surgeon. Topical antibiotic ointments have been found to not only decrease wound infections but also to provide a moist environment to promote epithelialization. Additional covering has not been found to decrease infection or improve healing. It has been noted that patients can get the wound wet after 12 hours with no increase in infection rates.¹⁰

Avulsions

Blunt trauma that produces a variable thickness flap of skin with or without underlying tissue constitutes an *avulsion*. In the acute setting, the simplest repair should be chosen. Questionably viable tissue should be preserved if excision would prevent primary closure. Closure should be

done with essentially no tension. Recruiting local tissue flaps in an acute situation often results in further ischemia and necrosis because of the disruption of local circulation. Wounds with extensive soft tissue loss should be treated with local wound care and serial debridement every 24 to 48 hours to establish viable wound edges. Final closure should only be performed after all remaining tissue is viable with no signs of infection.⁵

Many soft tissue defects in the face can be repaired with local rotation or transposition flaps. Large pedicled flaps and free tissue transfer may also be required in cases of significant loss, but rarely in the emergent setting. When skin grafting is required, special consideration must be given to the choice of donor site. Patient age and sex as well as graft thickness should be considered. Skin from hair-bearing sites should be avoided unless specifically indicated. For large, split-thickness grafts, the lateral thigh, buttock, anterior abdominal wall, medial thigh, and medial upper arm are reasonable choices. For smaller areas, when a full-thickness graft is required, the cervical, preauricular, and postauricular skin is preferred for its color match.²

Dog Bites

An estimated 4.7 million people sustain dog bites annually, with most occurring in the pediatric population, leading to more than 400,000 emergency department visits every year.³ Although bites can occur anywhere on the body, in children approximately 80% involve the head and neck because of a child's similar size in relation to the animal. The most common sites involved are the cheeks, lips, nose, and ears.¹¹ Although most of these bites can be treated with primary closure, the immense force of the bite can lead to more severe injuries, including complex soft tissue avulsions, facial fractures, or injuries to the airway or major blood vessels. In addition, even if the bite appears to be an isolated laceration or avulsion, there is often a crush injury component that may require further investigation. The severity of the injury increases in infants and younger children because of their smaller size and thinner tissues.¹² Regardless of the extent of the injury, medical management should include tetanus prophylaxis, prophylactic antibiotics, and rabies precautions (Fig. 15-1).

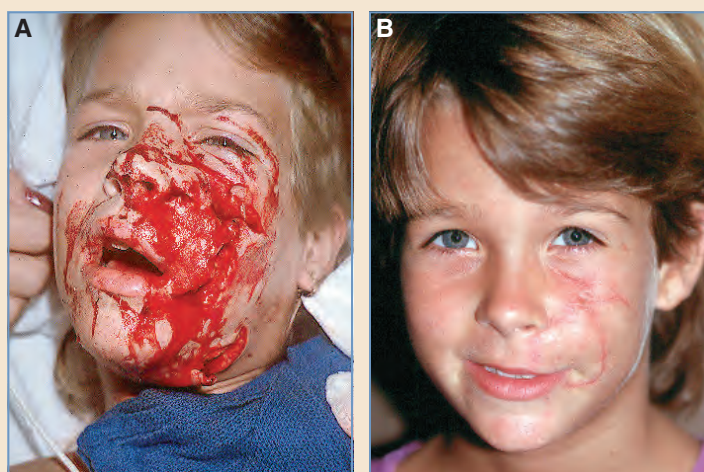


Fig. 15-1 A, A boy with a dog bite to the face. B, After careful wound care and closure, the patient appears to be doing very well. Although the scars are permanent, they will improve with time.

Wild animals must be considered rabid, and if possible, the brain of the offending animal should be examined for the presence of rabies antibodies. The patient should be treated with prophylactic rabbit immune globulin and human diploid cell rabies vaccine (HDCV) when indicated. If HDCV is not available, duck embryo vaccine may be substituted. If the bite is from a nonimmunized or questionably immunized domestic animal, the animal may need to be quarantined for 10 days, and if no illness or odd behavior is observed in the animal within this period, no treatment of the child is required.¹³

The rate of infection from dog bites ranges from 10% to 15%, with *Pasteurella multocida* and *Staphylococcus aureus* the two most common organisms involved.¹⁴ Antibiotic prophylaxis is commonly administered, especially in immunocompromised patients or those with contaminated wounds or wounds with delayed healing. Initial choices include oral amoxicillin or ampicillin with sulbactam for more severe cases requiring parenteral administration. For patients with a penicillin allergy, options include an extended-spectrum cephalosporin or trimethoprim-sulfamethoxazole plus clindamycin for anaerobic coverage. The length of antibiotic therapy can vary from 3 to 14 days, based on clinical judgment.¹⁵

On initial evaluation, the wound should be copiously irrigated with a mixture of normal saline solution and either a surgical preparation solution or an antibiotic solution. Cleansing should be followed by a thorough inspection to identify all injuries, with an awareness that both the upper and lower jaws of the dog may have caused damage. Debridement of facial wounds must always be minimal, but devitalized and shredded tissue should be removed to create a viable wound margin for closure. Irregular margins may be used to create a broken line for closure, reducing the need for unnecessary debridement of viable tissue. For cosmetic concerns, all dog bites to the face should be closed primarily if possible. Wounds with a very high possibility of infection or those more than 12 hours old should undergo delayed primary repair. In the event of significant soft tissue loss or underlying bone injury, the patient should be taken to the operating room for repair. Local tissue flaps and skin may be necessary for coverage, but these should not be placed over infected or contaminated wounds in the acute setting because of the increased risk of infection (Fig. 15-2).



Fig. 15-2 A, A child with a dog bite to the face treated with primary closure and local flaps. B, The patient shows normal animation of the face with residual scarring.

Thermal Burns

Burn injury is the fifth leading cause of accidental death in children, with thermal burns representing two thirds of all cases. Recent improvements in resuscitative measures, infection control, wound care, and nutritional measures have greatly increased survival.¹⁶ Although the pathophysiology and management principles of thermal burns are similar in children and adults, certain differences exist, including specifics of acute resuscitation, wound healing, derangements in facial growth, and increased emotional and psychosocial effects.¹⁶ A multidisciplinary approach with a well-thought-out plan is crucial to a successful functional and cosmetic outcome.

Thermal burns are caused by direct cellular destruction from thermal energy. The extent of damage is based on both the temperature and duration of exposure. Burns are classified based on the depth of injury. *First-degree burns* are limited to the epidermis and heal within 1 week with no residual scarring. *Second-degree burns* extend into the dermis and are further distinguished by the depth of dermal involvement (superficial versus deep). A superficial second-degree burn will heal within 1 to 2 weeks, but a deeper second-degree burn may take over 1 month to heal. *Third-degree burns* extend completely through the dermis into the subcutaneous tissues, causing loss of sensation as a result of local nerve injury. *Fourth-degree burns* are characterized by exposed muscle or bone. In general, deep second-degree burns and deeper burns require early excision and grafting. Children have thinner tissues than adults, which puts them at increased risk of more severe burns, and thus a child with a burn injury that appears similar to that of an adult may actually have more damage.¹⁷

First-degree burns are best treated conservatively, with localized care and bacitracin ointment. Superficial second-degree burns are also treated with antibiotic ointment (bacitracin or silver sulfadiazine) and subsequent coverage with a moist, semioclusive dressing. For auricular burns, mafenide acetate (Sulfamylon) is the preferred topical antibiotic, because it penetrates down to the cartilage and gives excellent coverage for *Pseudomonas* infection. For deeper burns, there are three phases of injury management: cleaning and debridement, tissue grafting, and reconstruction. The first phase lasts about 7 to 10 days and consists of topical antibiotics and local debridement to allow tissue demarcation before proceeding with excision and grafting. Skin substitutes, such as cadaveric allografts, xenografts, or a bioengineered membrane such as Integra, can be used for temporary coverage before definitive repair. Final reconstruction should start at least 1 year after injury to allow complete graft healing and scar maturation.

The healing process for burn injuries is often complicated by scar contractures, leading to multiple complications in the growing child. Periorbital contractures resulting in lid closure can be complicated by occlusion amblyopia, which is a loss of vision resulting from interruption of the developing visual axis. Perioral burns can cause microstomia, difficulties with speech and feeding, and alteration in dentition.¹⁸ Facial growth is also greatly influenced by symmetrical external forces from surrounding tissues, and alterations in these forces from scarring can lead to deformity. This is especially likely in the developing nose, which can be limited by surrounding contractures.

A discussion of all the reconstruction options for facial burns is beyond the scope of this chapter, but the key to a successful outcome is reconstruction based on specific facial aesthetic units. When multiple procedures are required, the recommended sequence for reconstruction is:

1. Perioral tissues
2. Neck, chin, or cheek
3. All remaining tissues

Treatment of pediatric burns remains a very difficult area for the plastic surgeon. Reconstruction is often a long-term process requiring a strong relationship between the surgeon and patient.

Electrical Burns

Electrical burns account for 4% to 7% of total burns, with many of them seen in young children who bite on electrical cords.¹⁹ The lower lip and lateral commissures are most often affected, but the teeth and supporting structures may also be involved. The extent of injury depends on the voltage, duration of contact, tissue resistance, and type of current. Household alternating current (AC) produces more damage than direct current (DC) of the same voltage.²⁰ The burn cauterizes local blood vessels and nerves, resulting in a painless necrotic lesion with minimal bleeding. Supportive care includes tetanus prophylaxis, administration of fluids, and antibiotic coverage. After initial eschar separation, secondary wound healing begins, usually resulting in significant scarring and microstomia.²¹ Early oral splinting is very important in this phase to prevent the formation of contractures. Oral burns can also affect craniofacial growth and development; therefore orthodontic appliances may be warranted in some patients to maintain growth.¹⁹ Reconstruction planning should begin once the healing process is complete. The hallmark of proper oral reconstruction is overcorrection to allow the inevitable postoperative wound contraction.

SPECIAL CONSIDERATIONS

Scalp Injuries

The scalp has an extensive vascular supply, which can produce significant blood loss when injured. Immediate application of a compression bandage will facilitate hemostasis before closure. The scalp consists of five layers: (1) skin, (2) subcutaneous tissue, (3) galeal aponeurosis (galea aponeurotica), (4) loose areolar tissue, and (5) periosteum. In general, most defects under 3 cm can be closed primarily, with or without galeal scoring.² Attempts should be made to close the scalp in layers, especially the strong galeal aponeurosis, which permits better skin approximation and helps prevent future development of a subgaleal hematoma. If time does not permit a layered closure, adequate control of hemorrhage and closure can be achieved with a single layer of running, interlocking sutures or staples. Any large arterial bleed identified should be suture ligated before closure. Associated skull fractures must always be suspected, and the surgeon should perform thorough palpation and inspection for any full-thickness scalp wound.

Eyebrow Injuries

Eyebrow lacerations should not prompt one to shave the remaining hair because of the unpredictable ability of the hair to grow back. The hairs of the eyebrow run oblique to the skin surface, so any additional incisions made for further debridement should follow the line of the eyebrows to limit hair loss. Lacerations should be closed meticulously in layers, including any muscles involved, with careful alignment of the superior and inferior margins. Approximation of the superior border takes priority, because misalignment in this area is most noticeable.³

Eyelid Injuries

In all traumatic eyelid injuries, the first priority is to rule out and avoid ocular injury. A topical anesthetic may be used to help with the examination and facilitate closure. A corneal shield may be used to avoid injury to the cornea. Full-thickness lacerations must be repaired by separately approximating each layer involved, beginning from deep (conjunctiva) to superficial (skin). A fine absorbable suture is used to carefully close the conjunctiva, avoiding corneal injury or irritation from the suture knots. The tarsal plate should be separately repaired with similar

absorbable suture. If the insertion of the levator aponeurosis or Müller's muscle is disrupted, it should be properly advanced to the tarsal plate at the time of repair to prevent later ptosis. The skin is closed with either absorbable or permanent interrupted or running sutures. Permanent sutures may be removed within the first week to minimize scarring. It is important to accurately align the gray line to restore the normal curvature of the eyelid margin. Any injury to the lower lid poses a greater risk of increased scleral show and ectropion formation. Although proper tissue realignment during repair decreases the risk of such complications, in some patients cartilage grafting, lateral canthopexy, or tarsal strip grafting is required to provide additional support.²²

Avulsion of the eyelid is a true surgical emergency. Immediate coverage is required to prevent further corneal damage, decreased visual acuity, and exposure keratopathy. On initial evaluation in the emergency department, an antibiotic patch should be placed over the cornea and the eye covered with a nonpermeable occlusive eye shield. Trapping moisture within the orbit helps to prevent dryness and further desiccation of the superficial corneal layers. An ophthalmologist should be consulted in all cases. Various reconstructive techniques have been described, with the common goal of replacing all three layers of the lid to restore function. The conjunctiva can be replaced with adjacent or cross-lid conjunctiva or conjunctival grafts consisting of oral or palatal mucosa. The tarsus is reconstructed by tarsal plate advancement or nonvascularized cartilage grafts, and skin is replaced with local tissue flaps or full-thickness skin grafts.²³

Lacrimal Duct Injuries

Lacrimal duct injuries occur in 16% of eyelid lacerations from either penetrating or blunt trauma.²⁴ Lacerations involving the medial canthal region are assumed to have an associated lacrimal duct injury. In most cases visual inspection of the wound will reveal the injury, but probing can be performed to confirm the diagnosis. Care must be taken to avoid exacerbating the injury during inspection and repair. If both ends of the ducts can be identified, the severed ends should be realigned, splinted internally, and repaired over a Silastic tube, which is generally left in place for 3 to 6 months. Some surgeons have elected not to directly repair the mucosal injury of the duct and have shown equivalent results repairing only the surrounding soft tissue to bolster the stent.²⁵

Nasal Injuries

The soft tissue of the nose includes the skin, subcutaneous fat, nasal muscles, cartilage, and endonasal mucosa, all of which are highly susceptible to injury as a result of the anterior projection of the nose from the face. All soft tissue injuries, including lacerations and avulsions of the nose, have the potential to cause both functional and cosmetic problems either as a result of tissue loss or poor healing. Intervention should be initiated as early as possible, beginning with judicious debridement, irrigation, and definitive repair. Full-thickness injuries should be repaired in layers. Nasal mucosa should be repaired with fine absorbable suture. Cartilage and skin should be approximated with fine nonabsorbable interrupted sutures, although in younger patients absorbable sutures or tissue adhesives may be more effective for skin closure.

The nose has multiple aesthetic units, including the tip, dorsum, paired alae, nasal sidewalls, and soft triangles. Each should be carefully addressed and reapproximated during the repair.²⁶ Significant cosmetic landmarks, such as the epidermal-mucosal junctions, nasal fold junctions, or alar rims, should be aligned first to decrease the likelihood of asymmetry.

Complications specific to the nose include nasal vestibular stenosis, with resultant airflow obstruction and chondromalacia of the nasal cartilage. Scarring and contracture of the alae cause obstruction after injury, and chondromalacia results from an untreated septal hematoma and can

eventually lead to a saddlenose deformity. If a septal hematoma is diagnosed, drainage should be performed immediately.²⁶

Cheek Injuries

The cheek is defined as the area bordered by the infraorbital region superiorly, the nasolabial fold medially, and the preauricular area laterally. The area consists of multiple important structures; therefore a comprehensive head and neck examination must be performed for all injuries. If the injury extends into the oral cavity, a layered closure is required, with fine absorbable sutures used for the mucosa and muscle layer and then either absorbable or nonabsorbable sutures for the skin, depending on the age of the child.³ Cheek lacerations may result in damage to the facial nerve; this will usually be evident during the head and neck examination. If the nerve is lacerated, a primary anastomosis should be performed during skin repair if the two ends are identified.

The parotid duct connects the parotid gland to the oral cavity, with the opening located adjacent to the maxillary second molar. If a duct injury is suspected, the orifice of Stensen's duct should be probed by injecting saline solution or dilute hydrogen peroxide through an angiocatheter, observing for communication with the wound. The use of methylene blue is discouraged, because it can cause discoloration of the tissues. If injury is confirmed, the proximal end can be identified by the expression of saliva from the gland. A catheter is then positioned through the laceration via Stensen's duct, and the duct is repaired over the catheter.²⁷

Lip Injuries

In evaluating facial appearance, the human eye is initially drawn to the lips and eyes, emphasizing the importance of these structures.²⁸ Particular care must be taken to align three important labial structures: the vermilion border, the orbicularis oris muscle, and the red line. The vermilion border and red line can be marked with methylene blue or tacking sutures to help facilitate meticulous alignment. This is especially helpful if performed before infiltration of local anesthetic, which can distort these areas.

The *vermilion border* (white roll) marks the border between the vermilion and the cutaneous skin. This structure should be the first area to be precisely approximated during closure. Two sutures, one on each side of the border, should be placed to align opposing tattoo marks or tacking sutures. Failure to properly align this structure is usually apparent from several feet away, even with a 1 mm difference. The second area to be approximated should be the red line, which forms the junction between the wet and dry mucosa of the lip. Failure to properly realign this structure results in a vertical distortion of the lip vermilion and chapping of the lip from eversion of the wet mucosa into the dry area. In general, sutures placed in the mucosa should be absorbable, and cutaneous skin sutures can be either fast-absorbable or nonabsorbable, depending on the age of the child. An interrupted or figure-of-eight absorbable suture should be used to approximate the orbicularis oris muscle. Poor reparative efforts in this area will lead to both abnormal lip movement and muscle bulging lateral to the bite area.²⁸ After repair, patients should be instructed to rinse their mouths five times daily with an antimicrobial mouthwash.

External Ear Injuries

Because of its exposed position on the head, the external ear is commonly involved in traumatic injuries. The rich vasculature of the ear allows excellent healing if properly treated, even with severe injuries. After adequate debridement of devitalized or jagged tissue, full-thickness ear lacerations should be repaired in a three-layer closure with either fine absorbable or nonabsorbable sutures for the cartilage and skin (anterior and posterior). If there is a large area of exposed cartilage without available skin coverage, this cartilage can be preserved within a subcutaneous pocket in the mastoid region for future reconstructive efforts.²⁹

Ear cartilage is very susceptible to deformity from hematoma, and thus meticulous hemostasis should be performed before closure. *Cauliflower ear* is the result of recurrent blunt trauma, causing formation of subperichondrial hematomas. These collections create a chondroinductive matrix for chondroblasts contained within the elevated perichondrium. Chondroblasts then form ectopic cartilage within the area, inducing significant deformity. Prevention includes rapid incision and drainage of any auricular hematomas. Meticulous hemostasis must be achieved, followed by primary closure. In a large hematoma, a suction drain may be left in place for several days.³⁰ If the patient has already developed this condition, the heterotopic cartilage can be directly excised through multiple approaches.³¹ A compression dressing is then applied to maintain proper tissue contour and prevent further bleeding.

Complete avulsion or amputation of the ear requires immediate microvascular repair. When successfully completed, repair typically results in full restoration of the external ear structure (see Chapter 33). Delayed management of an avulsed ear often requires total ear reconstruction. Partial avulsions may be treated with sharp debridement, followed by local tissue advancement flaps. The opposite ear or costal cartilage can be used. More significant defects require harvesting cartilage from the opposite ear and covering the free cartilage graft with a temporal parietal flap and full-thickness skin graft.

Nerve Injuries

The facial nerve is the motor nerve that is most vulnerable to both blunt and penetrating facial trauma and results in serious functional and aesthetic defects. An early, accurate examination of facial animation should be performed after any injury.³² So that all of the critical branches of the nerve can be assessed, patients should be asked to raise their eyebrows (frontal branch), close their eyes (zygomatic), smile (buccal), and pucker their lips (marginal mandibular). In penetrating injuries with transection of all or portions of the nerve, deficits will be seen immediately; however, in blunt trauma there may be a partial or delayed effect. Transection injuries that occur anterior to the lateral canthus are often only observed because of the sufficient crossover from the opposite side. If the injury is posterior to the lateral canthus, early identification and repair by direct coaptation or cable grafts will achieve the best results.³³ If there is substantial nerve loss or a grossly infected field that precludes immediate repair, the nerve ends should be identified and tagged for later repair (Fig. 15-3).

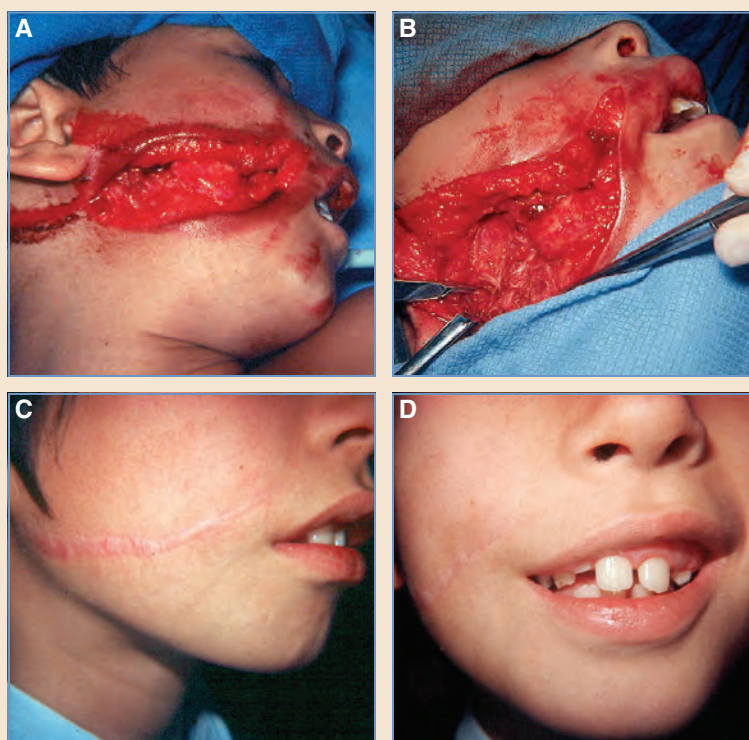


Fig. 15-3 A patient with a severe injury of the facial area, including the facial nerve. **A**, A large open wound of the cheek. **B**, The facial nerve is exposed and repaired. **C**, Resultant scar. **D**, Partial return of animation 1 year later.

Sensory nerves, such as the supraorbital, supratrochlear, infraorbital, and mental nerves, may be injured by trauma; however, the resultant hypesthesia typically causes only minimal long-term effects.

MANAGEMENT OF COMPLICATIONS

Common complications in pediatric patients are related to poor scarring (widespread, hypertrophic, hypopigmented, or hyperpigmented) or keloid formation, but hematomas and wound infections also occur and require immediate attention. Children often heal with hypertrophic scars that will flatten and fade over time. It can take more than 1 year for the redness to fade and the scar to soften (see Fig. 15-3, *C* and *D*). Because of this, revisions, when necessary, should be delayed for a minimum of 6 months, if not more than 1 year. Initial scar treatment with a pulsed-dye laser or dermabrasion may be sufficient, and the anticipated secondary surgery may become unnecessary.³ Conversely, there is no need to wait until adulthood to repair such scars, which may carry significant psychological implications and potentially have deleterious effects on development. If the patient has a history of poor scarring, techniques for prevention include triamcinolone acetonide (Kenalog) injections, pressure dressings, and limiting skin stretching along the scar. To limit skin stretching during healing and thereby facilitate appropriate wound resting, wounds should be



Fig. 15-4 An infected facial wound. Traumatic injuries are considered contaminated, with an increased risk of infection. For treatment of wound infections, sutures should be removed and the wound allowed to close by secondary intention.

covered by fixable materials, including tape, bandages, garments, or silicone gel sheets.³⁴ In addition, the use of topical steroids can help alleviate the itching associated with keloids.³⁴

Prevention is the best treatment for infections and hematomas. The use of sterile technique, administration of antibiotics when necessary, and application of proper hemostatic measures can be very effective. For acute wound infections, it is often necessary to open a portion of the wound to allow drainage of any accumulated fluid. Wound cultures should be taken, and the patient is then started on empirical antibiotics until the results of cultures and sensitivities return. The wound should remain open until the infection has resolved and can then be reclosed or allowed to heal by secondary intention, depending on the circumstances (Fig. 15-4).

Stable hematomas can be observed and treated with warm compresses, but expanding hematomas require intervention. The wound should be reopened and explored for active bleeding. Once hemostasis is achieved, the wound can be reclosed and covered with a pressure dressing for added prevention.

CONCLUSION

Facial soft tissue trauma in the pediatric population is very common and requires both proper management of the injury and appropriate handling of the patient's and parents' concerns. Early assessment and treatment of facial injuries is the key to preventing immediate complications and ultimately achieving a successful outcome. Insufficient or delayed treatment can lead to long-term disfigurement, which can have a significant impact on the patient's psychological development. Considerable attention should be given to preparing the patient and parents for the healing process and the possibility of complications, with the need for future revisions. In addition to detailed documentation of location, size, depth, and associated injuries, photographs are very important to help show progress. In the acute stages of treatment, children benefit from a more conservative approach with the simplest possible repair, with more complex procedures reserved for secondary repair. Primary closure can usually be achieved with copious irrigation, judicious debridement, and meticulous reapproximation. In cases of significant tissue loss that prevents primary closure, the wound should be optimized before reconstruction with grafts or tissue flaps.

Ultimately, the management of facial soft tissue trauma is similar for children and adults—both require mastery of the full spectrum of reconstructive techniques to restore and preserve function while achieving optimal cosmetic results.

KEY POINTS

- Evaluation of all patients with head and neck trauma should begin with the ABCs to confirm that the patient has an adequate airway, is breathing appropriately, and is maintaining systemic circulation.
- Children are often frightened of medical personnel, and initial contact should focus on gaining their trust, reassuring them, and achieving mutual cooperation.
- For all major injuries, a local anesthetic should be administered before wound cleansing and exploration to prevent unnecessary pain during the process.
- Most soft tissue injuries in children can be managed in the emergency department or office with a local anesthetic, with or without sedation, but the use of general anesthesia in the operating room may be appropriate in special circumstances.
- Primary closure can usually be achieved with copious irrigation, judicious debridement, and the use of systemic antibiotics for highly contaminated wounds, especially human or dog bites.
- For an acute traumatic injury, children benefit from a more conservative approach, using the simplest possible repair and reserving more complex procedures for secondary repair.

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16

Injections, Infiltrations, and Envenomations

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any injection, infiltration, and envenomation injuries appear innocuous at initial presentation, but they can result in devastating consequences if they are not promptly recognized and effectively managed. Infiltration injuries typically involve the extremities, most commonly the hand and forearm, whereas injection injuries usually involve the nondominant hand. A variety of land and sea envenomations can result in serious injury, loss of function, and death. Many envenomation injuries can be managed expectantly with local measures, although some may require surgical and emergent intervention, including antivenin.

HIGH-PRESSURE INJECTION INJURIES

High-pressure injection injuries are rare in children and frequently present as an innocuous injury, but they can result in severe morbidity, including loss of function and the need for amputation. Patients frequently delay seeking treatment. The initial assessment and treatment by medical staff may be inadequate and the destructive potential of the injury underappreciated. Significant functional limitations and amputation rates of up to 48% have been reported.¹ Injection injuries typically result from high-pressure sprayers affecting the nondominant hand. Pressures generated from sprayers can range from 3000 to 69,000 psi.² The pressure necessary to penetrate skin is only 100 psi, and higher pressures can penetrate skin without direct contact.^{3,4}

Factors that affect the magnitude of injury include the interval length from injury to treatment, the type and toxicity of the injected material, the pressure/velocity of the injection, the amount injected and its temperature, the anatomy and distensibility of the involved tissues, ma-

terial viscosity and spread, and secondary infection.⁵ Tissue destruction results not only from the mechanical trauma of a high-pressure injection, but, more importantly, from the toxicity of the injected material.

Commonly injected industrial agents include grease, fuel, hydraulic fluid, paint, and solvents. Grease and hydraulic fluid are more viscous and spread relatively less than other agents. These injuries may result in fibrosis, scarring, chronic fistula, and progressive loss of function. Paints, thinners, and solvents have a lower viscosity and spread more easily. These agents tend to create a much more vigorous inflammatory response than grease or hydraulic fluid. A negative synergistic effect may result from the mechanical and chemical injuries, leading to severe edema and vascular compromise. Unless injuries are recognized and treated early, tissue necrosis can result, necessitating amputation.

Authors agree that the degree of tissue damage correlates closely with the substance injected.^{6,7} Lin et al⁶ demonstrated that paint in a rat model caused significantly more tissue damage than a saline solution at similar pressures. Gutowski et al⁸ described five patients who had high-pressure injection injuries with dry-cleaning solvents that included isoparaffins, methoxypropanolol, and dichloroethane. These substances can have relatively low systemic toxicity but severe local toxicity with tissue necrosis.^{9,10}

High-pressure injection injuries with water or gases, such as air, tend to be significantly less damaging, because these materials have low toxicity. Other substances have also been implicated in high-pressure injection injuries (Box 16-1).

Anatomy affects the degree of spread and injury as well as treatment. The spread of injection depends not only on the pressure of injection and viscosity of material, but also on the resistance of the tissue. For example, the dorsum of the hand provides significantly less resistance to a spreading substance than the palm. Injection over the flexor tendon sheath may result in spread along the flexor tendon sheath and into the hand and forearm. Injection injury to the relatively nondistensible and small space of the finger pulp can result in severe mechanical trauma and ischemia despite limited spread of injection material.

High-pressure injection injuries require prompt treatment to prevent significant morbidity, loss of function, and amputation. The mechanism of injury and the material injected must be identified. Radiographic studies can help delineate the spread of material. Paint, grease, and other agents are radiopaque to varying degrees. The extent of water spread can be determined by the extent of subcutaneous emphysema.

The extremity should be kept elevated, and all patients should receive tetanus prophylaxis and intravenous, broad-spectrum prophylactic antibiotics. The use of systemic steroids may modulate the inflammatory response; however, their use is controversial and is not particularly supported by the existing literature.

Box 16-1 Substances Associated With High-Pressure Injection Injuries

Air	Grease	Oil
Cement	Hydraulic fluid	Paint thinner
Dry-cleaning solvent	Molten metal	Turpentine
Freon	Molten plastic	Water

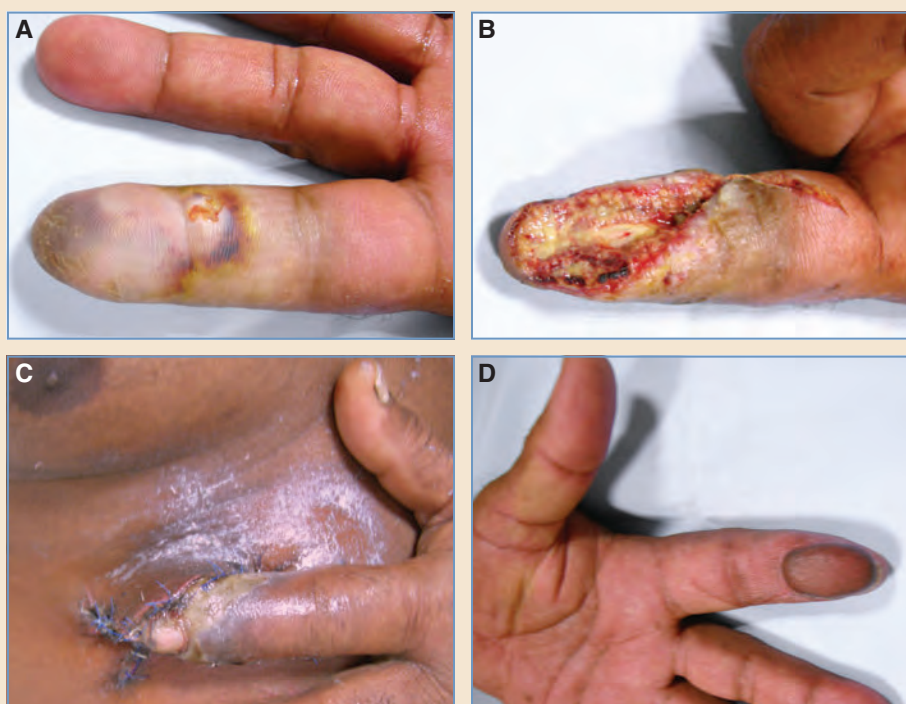


Fig. 16-1 A, This patient had a left index finger small puncture wound just proximal to the distal interphalangeal joint crease with surrounding induration, loss of sensation, and compromised vascularity of the volar skin distal to the proximal interphalangeal joint crease. B, The wound was explored with a Bruner zigzag incision. The resultant volar defect with exposed tendons is shown after the final wound debridement. C, The volar defect over the left index finger was covered by a superior-based abdominal flap. D, The appearance 3 months postoperatively. (Courtesy of Sanjay Saraf, MD.)

All cases of high-pressure injection injuries, with the possible exception of air, other gases, and water, should be considered surgical emergencies. Wide surgical exploration, debridement, and irrigation with removal of foreign material are essential. A tourniquet may be used, but Es-march exsanguination is discouraged to prevent further proximal dispersion of injected material. Bruner or midlateral incisions can be used for injection injuries to the volar surface of the hand or fingers, and the flexor tendon sheath should be explored¹¹ (Fig. 16-1). Repeat surgical exploration, debridement, and irrigation are frequently necessary. Open wound management may be necessary, followed by delayed closure with good outcomes.¹² Early physical therapy to prevent loss of function is of critical importance, and patients should be advised at the outset of the significant potential loss of function or even amputation. High-pressure injection injuries should be considered a surgical emergency, and any delay in surgical intervention results in poorer outcomes.^{13,14}

INFILTRATION INJURIES

Infiltration injuries are surprisingly common especially in children, with a reported incidence of 11% of all NICU patients^{15,16} (Fig. 16-2). Neonates and infants have small and delicate veins, and unlike older patients, are unable to verbalize pain at an IV site. The smaller the patient, the



Fig. 16-2 IV infiltration injuries with full-thickness skin loss of the distal radial surface.

greater the potential injury from infiltration. Despite these difficulties, most infiltration injuries are preventable. Fastidious nursing care with careful monitoring, automated IV pumps with sensors, and proper dressing and taping significantly reduce infiltration injuries. *Infiltration* and *extravasation* are often used interchangeably. Some authors refer to infiltration as leakage of non-toxic fluid such as normal saline solution, whereas extravasation is reserved for leakage of toxic fluids such as osmotically active agents, ischemia-inducing agents, and chemotherapeutic agents.¹⁷ Other authors have described extravasation as leakage of vesicant agents, which results in blistering and tissue necrosis, and infiltration as leakage of irritants that may cause redness, pain, and inflammation, but rarely tissue necrosis.^{18,19}

Many factors contribute to the morbidity of these injuries, including the site of infiltration, the size of the patient, the amount of fluid leaked into the tissue, the toxicity of the leaked fluid, the general medical condition of the patient, and management. Hospitals have developed and adapted a variety of grading scales that help to effectively communicate the severity of the infiltration and guide treatment.²⁰⁻²²

When these injuries occur in areas with little subcutaneous fat, such as the dorsum of the hand or foot, structures can be exposed, even with minimal tissue loss. The mechanism of tissue damage caused by extravasated agents can be classified as osmotic, ischemic, direct cellular toxicity, and mechanical compression. Secondary infection can also contribute to tissue damage. The agent and its mechanism of tissue damage determine the recommended treatment.

Osmotically Active Agents

Osmotically active agents include hypertonic dextrose solutions, parenteral nutrition solutions, sodium bicarbonate, radiographic contrast dyes, and solutions containing calcium and potassium. These substances can cause tissue damage by creating local osmotic imbalances that can result in cell death by disrupting cell membrane transport or intracellular dehydration. Because solutions containing calcium and potassium are also slightly acidic and hypertonic, they are capable of precipitating proteins, which also results in cell death.²³

Ischemia-Inducing Agents

Ischemia-inducing agents include those with alpha-adrenergic properties, such as epinephrine and dopamine. Certain cations, such as potassium and calcium, may cause ischemia through depolarization and contraction of smooth muscle sphincters. Dopamine is arguably the most common vasoactive drug in extravasation injuries.²⁴ Treatment options used for osmotically active agent extravasation have also been employed for vasoactive agents. Casanova et al²⁴ described using hyaluronidase in an attempt to disperse and dilute the vasoactive agent. Gault²⁵ described a saline flush technique that has been used in cases of dopamine extravasation, but it remains somewhat controversial.²⁵ The best results were obtained when treatment was initiated within 6 hours of extravasation. Other authors have advocated specific antidotes, such as phentolamine and topical nitroglycerin for dopamine extravasations²⁶⁻²⁸ (Fig. 16-3).

Chemotherapeutic Agents

Chemotherapeutic agent extravasation injuries arguably result in the most significant morbidity and can be categorized as vesicants, nonvesicants, or irritants (Box 16-2). Vesicants can be further divided into DNA-binding and DNA-nonbinding (Box 16-3). DNA-binding vesicants bind cellular nucleic acid, resulting in cell death and necrosis. Toxic complexes are released from dead cells and taken up by adjacent, healthy cells, leading to further tissue necrosis and progressive tissue loss over weeks to months.¹⁷ DNA-nonbinding vesicants, including plant alkaloids, are less cellularly toxic with less resulting tissue damage and tend to remain more localized.



Fig. 16-3 A dopamine infiltration injury.

Box 16-2 Chemotherapeutic Agents

Vesicants

Dactinomycin	Mitomycin C
Daunorubicin	Nitrogen mustard
Docetaxel	Paclitaxel
Doxorubicin	Vinblastine
Epirubicin	Vincristine
Idarubicin	

Nonvesicants (irritants)

Bleomycin	Ifosfamide
Caroplatin	Irinotecan
Cisplatin	Melphalan
Cyclophosphamide	Oxaliplatin
Etoposide	Thiopental
5-Fluorouracil	

Box 16-3 Vesicants**DNA-Binding**

Daunorubicin	Mitomycin C
Doxorubicin	Bendamustine
Idarubicin	Mechlorethamine
Dactinomycin	

DNA-Nonbinding

Vinorelbine	Docetaxel
Vinblastine	Paclitaxel
Vindesine	

Treatment will depend on the type of chemotherapeutic agent. Dexarazoxane is recommended for anthracycline DNA-binding agents, including daunorubicin, doxorubicin, epirubicin, and idarubicin. Its mechanism of action includes binding iron, preventing the formation of free radicals, and binding and inactivating DNA topoisomerase II; it should be injected within 6 hours and away from the site of extravasation.²⁹ DNA-nonbinding vesicants and irritants can be treated with heat or topical nitroglycerin to promote vasodilation and dispersion of the drug. Hyaluronidase can be used to break down hyaluronic acid, promoting diffusion and absorption of the drug. Other treatment options include infusion of saline solution to dilute the drug, with or without subsequent liposuction techniques to extract the offending drug. If such an approach is considered, it should be done within 6 hours of the extravasation. Sodium thiosulfate has been recommended for mechlorethamine and bendamustine extravasation and is thought to neutralize the formation of free radicals and subsequent tissue necrosis.³⁰

A variety of other antidotes, including vitamin C, heparin fractions, dexamethasone, glucosteroids, alpha-tocopherol, dapsone, and dimethyl sulfoxide have been advocated for the treatment of chemotherapeutic extravasations; the possible benefits suggested based on animal studies are unproven in humans.³¹⁻³⁶

In most cases, successful management of infiltration injuries requires careful monitoring and early detection, discontinuation of the infusion, administration of the appropriate antidote, and observation, reserving the possible need for debridement and skin graft or flap coverage for tissue necrosis or evolving lesions.

ENVENOMATIONS

Many species of snake, reptile, arachnid, insect, and sea creature have the ability to deliver noxious substances through bites, stings, or other means for hunting prey and/or for self-defense. Envenomation of humans is almost always defensive in nature. Many of these injuries can be managed expectantly with local measures, but others can be lethal and require emergent intervention and antivenin.

Snakes

Approximately 4000 to 6000 snakebite envenomation injuries occur in the United States annually, resulting in approximately five deaths.^{37,38} Most venomous snakes in the United States include pit vipers and coral snakes, although increasingly, pet enthusiasts are introducing other venomous exotic snakes. Common pit vipers in the United States include rattlesnakes and copperheads.

Most snakebites occur on the lower extremities as a defensive reaction of the snake. Once a snakebite has occurred, the victim should be moved a safe distance from the snake, and any restrictive jewelry and clothing should be removed. If an extremity has been bitten, then it should be immobilized. Tourniquets should not be used, because they can exasperate ischemia and toxin-mediated injury.³⁹ Pressure immobilization can be used in an attempt to limit spread of the venom. Typically, the involved extremity is immobilized with a splint, and an elastic bandage is applied from proximal above the snakebite to distal.⁴⁰ Incision and oral suctioning should not be performed, because it is of no benefit and can introduce oral bacteria into the wound.³⁹ The patient should be kept calm and immobilized and immediately transported to a hospital.

Signs and symptoms of snakebite envenomation range from local pain, swelling, and erythema to systemic manifestations of nausea, vomiting, paresthesias, confusion, muscle tenderness, fasciculations, respiratory paralysis, and death. On arrival at the hospital, patients are evaluated. This includes laboratory studies, and hemodynamic and respiratory support are provided if necessary. The poison control center is contacted, and the appropriate antivenin given if available. Local wound care is performed and a fasciotomy considered if compartment syndrome is evident.

Arachnids

Arachnidae is a class of arthropod that includes spiders, ticks, mites, and scorpions. It includes more than 75,000 species worldwide, 20,000 of which are spiders.⁴¹ Approximately 18,000 species of spiders are venomous. Most cause only local irritation, whereas some are capable of inflicting serious local and systemic damage.

Black Widow

The black widow (genus *Latrodectus*) is probably the most feared spider in the United States. Five species have been found throughout the country, more commonly in the southern states. They are identified by their typical glossy black body and orange hourglass marking on the ventral surface of the abdomen, although only one species carries these specific markings; some carry various markings on their dorsal side. The female spider of this species is typically 1 to 1.5 cm long (three times larger than the male) and is responsible for most bites to humans. Bites usually occur between April and October, because the female hibernates during the winter.

Black widow venom is considered one of the most potent secreted by any animal, and an adult has enough venom to kill a 30-pound child.⁴² The venom contains a neurotoxin, alpha-latrotoxin, that causes massive release of acetylcholine and norepinephrine at the neuromuscular junction, eventually leading to depletion of transmitters and blockage of further signals.⁴³

The bite often causes mild to moderate pain and leaves a wheal and flare. A cramping pain begins at the site of the bite and gradually spreads to the larger muscles, including the abdominal wall. Severe abdominal cramping and pain are the hallmark symptoms of black widow spider bites. Fine muscle fasciculations can sometimes be seen. Nausea, vomiting, weakness, and anxiety are also common. Paralysis and respiratory arrest can occur in children. Severe hypertension is possible.⁴⁴ Treatment includes pain relief with opiates, muscle relaxants, and antivenin if any neuromuscular symptoms are present. Muscle relaxation can be achieved through a variety of measures. Calcium gluconate 10 ml of 10% solution may cause significant pain relief and perhaps is diagnostic, but cardiac monitoring is required with its administration. Valium may also be given in appropriate doses. A horse serum antivenin is available that has a dose of 1 vial for both children and adults, relieving symptoms in 1 to 3 hours, although serum sickness is possible.⁴⁴

Recluse

Recluse spiders (genus *Loxosceles*) are found most commonly in the southern United States, although they are not confined to these areas. Of the 13 species known, the brown recluse (*L. reclusa*) is the most prevalent. It is often called the *violin* or *fiddleback spider* because of the characteristic violin-like marking on the anterodorsal cephalothorax. Of all spider bites, plastic surgeons are most often consulted for that of the brown recluse because of the potential for soft tissue necrosis.

KEY POINTS

- Early recognition and appropriate treatment are paramount for treating injuries secondary to injection, infiltration, and envenomation.
- Improper or delayed treatment may result in loss of function or the need for amputation.
- Knowledge of medical and surgical treatments should be part of the armamentarium of every plastic surgeon to help improve outcomes for these injuries.

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Part II



Congenital and Acquired Deformities of the Facial Skeleton and Soft Tissues of the Head and Neck

17

Psychological Issues Affecting the Treatment of Children and Adolescents With Cleft and Craniofacial Conditions

Kathleen A. Kapp-Simon



Children born with cleft lip and palate (CL/P) and other craniofacial conditions (CFCs) face numerous psychological challenges throughout their lives. This chapter identifies psychological issues that affect adjustment, including differences in family adaptation and child temperament, social skills, and cognitive abilities. Specific issues related to surgical intervention are discussed, along with methods of promoting healthy psychological adjustment in children undergoing treatment for CL/P and other CFCs.

The psychological care of children born with a CL/P and other CFCs is complex and ongoing, beginning at birth and for most children, continuing to young adulthood. This care encompasses issues related to family adaptation, coping with surgeries and other therapeutic interventions, self-esteem, social competence, emotional and behavioral adjustment, cognitive functioning and school achievement, and ultimate integration into society as a fully functioning adult. The plastic-craniofacial surgeon is in a unique position with regard to a number of these issues. The surgeon may be one of the first persons with whom a family speaks who is knowledgeable about the child's condition. In addition, the surgeon has the power to bring about major changes in the child's appearance and functioning. Thus the surgeon's initial presentation about the child's condition and its possible effect on a child's life has the potential for long-term influence. In addition, the surgeon, in conjunction with the cleft-craniofacial team, will see a child and his family

through the many years of habilitation. Consequently, the surgeon will often hear about a child's problems or family concerns in areas beyond his or her expertise.

We will discuss psychological issues as they relate to the clinical care of children with clefts or other CFCs. The initial focus is on the parents and their adjustment to their child's birth. This section is followed by a general discussion of issues related to emotional, behavioral, social, and academic adjustments that affect treatment decisions at various ages in a child's life. Finally, a framework for surgical decision-making during adolescence is proposed.

PARENTAL ADJUSTMENT

Whether diagnosis occurs prenatally or in the delivery room, most parents are unprepared for the news that their child has a CL/P or CFC.¹ Thus they must cope with feelings of shock, loss, grief, and worry as they try to accomplish important tasks that may be as variable as learning to feed their infant, managing their infant's airway, making decisions about immediate treatment priorities, and seeking information about long-term care.² An early referral to both a cleft-craniofacial team and to a local support network or national support program such as The Cleft Palate Foundation (<http://www.cleftline.org>), Ameriface (<http://www.ameriface.org>), or the Children's Craniofacial Association (<http://www.CCAkids.org>), will provide parents with critical information and options for emotional support. There are also online support groups that are specific to the diagnosis. For example, there is an online support group for parents of children with CL/P on Facebook. However, many of these diagnosis-specific groups have short lives; thus referring professionals should be aware that it is not uncommon for websites to be out of date.

Parents are influenced by the attitude that medical professionals display toward their child. While the potential of an individual child can never be known fully at birth, an optimistic presentation of the child's future—with assurances that much can be done to both improve appearance (when appropriate) and foster optimal development—will provide the reassurance that anxious parents need. During clinic interviews with families of older children, the parents have often commented that early encouragement from a medical professional to think of their infant as a “normal baby” enabled them to get past the initial feelings of grief and loss that are so typical.

It is also important to recognize that families who have a child with a CL/P or CFC have personal and family histories that they bring to coping with this new situation. A parent's level of prebirth adaptation and general emotional health as well as the level of family cohesion will influence a parent's ability to cope with the child's birth and will have an impact on long-term family and child adaptation.³ The initial process of adaptation can be challenging, because each parent may have different emotional reactions. One of the parents may be feeling very sad or depressed, whereas the other is angrily searching for someone or something to blame. The degree to which parents are able to validate each other's experiences and coping process will influence the family's long-term adaptation and their child's mental health.

Parental competence is enhanced when parents recognize their differing strengths. One parent may be better equipped to seek out information about available medical/therapeutic treatments, whereas the other parent may do better supporting the young child through surgery. Professionals working with families can recognize and encourage these individual strengths. We do know that when parents believe that they can meet their child's needs, feel satisfaction in their role as parents, and realistically acknowledge the stresses of coping with the issues of raising a child with more complex medical needs, the parent-child relationship can be enhanced in important ways.

Despite the extensive evidence documenting the distress of the initial period following the birth of a child with a chronic medical condition such as CL/P or CFC, most families cope adequately and provide an emotionally healthy environment for their child. However, children are

not only affected by their families but also by societal reactions. There is considerable evidence to suggest that individuals with facial differences must negotiate a plethora of personal challenges to self-esteem and social acceptance after they leave the protective environment of the family.^{4,5} Although most individuals with CL/P or CFC are ultimately successful in coping with these difficulties and mature into productive and contributing members of society,⁶ there can be many sources of stress during the childhood years.

BEHAVIORAL, EMOTIONAL, AND SOCIAL ISSUES

Psychological adjustment in childhood is based on a child's behavioral competencies, emotional adjustment, and social adaptation. Behavior and emotional adjustment encompasses the ability to both internalize and externalize problems. Research has shown that children with CL/P and CFCs are at greater risk of exhibiting personal inhibition, including withdrawal, shyness, separation anxiety, and occasionally depression.⁷⁻¹⁰ There is recent evidence that children 4 to 9 years of age with oral clefts screen positive for separation anxiety disorder significantly more frequently than children without oral clefts (24% screened positive compared with a general population rate of 3% to 5%).¹⁰ However, there are also reports of more frequent acting-out behaviors, which may be age dependent. In one of the only longitudinal studies assessing behavior in children with CL/P, higher rates of acting-out behaviors were identified for boys at 6 and 7 years of age, which decreased over time. Girls also displayed acting-out behaviors, but not until 11 to 12 years of age. Internalizing problems was higher than the norms for both sexes; however, inhibition increased for girls as they got older, whereas it remained fairly constant for boys.¹¹

Issues related to self-concept have been intuitively linked to facial appearance, with parents, surgeons, and other team professionals assuming that improved facial appearance results in more positive global self-perception by the child or adolescent and greater social acceptability from peers. As with many commonly held perceptions, there seems to be some truth to this linkage. Children frequently blame social difficulties on facial differences.^{12,13} Peer teasing and victimization of children with CFC are often attributed to the presence of facial disfigurement.⁵ Recent quality-of-life research has found that even adolescents perceive that they are teased and made to feel unwelcome by peers frequently and that facial appearance has a strong influence on social relationships.¹⁴ Teasing from peers is not a benign experience for children. Peer teasing is associated with poorer psychological functioning, including increased symptoms of depression, anxiety, behavior problems, and dissatisfaction with appearance.^{15,16} Children seen in cleft and craniofacial clinics should be routinely screened to assess the frequency and type of peer teasing that they are experiencing. Schoolwide interventions are often needed to counter the negative environment that permits peer teasing at school.¹⁷

In addition, children with facial differences will benefit from being taught strategies to enhance their ability to interact appropriately with their peers. Social skills are specific positive behaviors that can enhance social interaction. Children with better social skills are reported to have fewer behavior or emotional problems and are less likely to be socially inhibited or withdrawn.^{18,19} Social skills, such as making eye contact, maintaining appropriate social distance, entering and maintaining conversations, managing social anxiety, and expressing empathy are all skills that can be taught.²⁰ Preliminary research findings indicate that children who are taught specific social skills can significantly increase the frequency with which they initiate contact with their peers and the frequency with which these contacts lead to more social interaction.²¹ Recently, the Center for Appearance Research in Bristol, United Kingdom, has launched research evaluating an on-line program for individuals with facial differences that incorporates current knowledge about social interaction, social anxiety, and cognitive behavioral interventions into a seven-session on-line teaching program: www.yfaceit.co.uk.²² A similar program for adults has been successful in decreasing anxiety and depression and increasing positive adjustment.²³

COGNITIVE FUNCTIONING

There is considerable research to support a higher risk of learning disorders in children with CL/P and other CFCs.²⁴ Thirty percent of children with CL/P (compared with 5% to 15% of the general population) experience specific reading disabilities,²⁵ and children with other types of CFC may experience learning problems resulting from hearing loss, reading disabilities, problems with attention, or nonverbal learning disabilities.²⁶ Children with CL/P access special education services at a more frequent rate.²⁷ Population-based studies suggest that as a group, children with CL/P lag behind their peers in most academic areas, even though individuals with CL/P may be very successful.²⁸⁻³⁰ There is growing evidence of a neurobiologic basis for at least some of the learning challenges that many children with CL/P experience. The research by Nopoulos and colleagues³¹⁻³⁷ has identified numerous changes in brain morphology and shape that are associated with cognitive functioning and specific aspects of academic achievement in individuals with CL/P. These authors have repeatedly asserted that individuals with CL/P are at risk for having a neurodevelopmental disorder that has an impact on both learning and social behavior.³⁸

Learning problems become most evident during the early elementary school years, particularly as demands for reading competency emerge. Consequently, a significant proportion of the young children seen by cleft and craniofacial teams are coping with multiple risks to adjustment at a very young age. Integration into the peer group is often more difficult for children who have learning problems, especially if they also have facial differences and behavior problems. Learning problems can add to a child's feeling of lowered self-confidence.

School districts do not always recognize that a child with CL/P or CFC has a specific learning disability. A child who is inhibited and has the stigma of a facial difference may be viewed as less intelligent by a teacher, and the true processing deficits that account for the child's learning difficulties may not be identified.³⁹ At the other extreme, a child who acts out in the presence of learning problems and social difficulties may be diagnosed with an attention deficit disorder, and the underlying learning disorder may not be properly identified or treated.⁴⁰ Current practices to screen academic progress for all children on a regular basis as part of the new educational initiative called Response to Intervention (RtI) has increased the probability that children will be receiving some interventions at an earlier age; however, it has also meant that children who are struggling may not receive appropriate diagnostic assessments that would identify learning disabilities in a timely fashion. As a consequence, the required intensity of early intervention may be delayed unnecessarily.

Cleft teams are in a unique position to educate families about the possibility of learning disorders in their children. Neurodevelopmental screenings should be provided, beginning in infancy. Children identified at risk should be referred for early intervention services. Preschool screening should include an assessment of phonological awareness, because weakness in this area is an early indicator of potential learning challenges. Academic progress should be evaluated during the early elementary school years, ideally by the cleft or craniofacial team. Children who are not keeping up academically with peers should be evaluated for specific learning disabilities. If resources are not available within the cleft team, parents should be instructed in their legal rights to an assessment through their school district (see <http://idea.ed.gov/> for further information on special education rights), or they should be referred to the psychology departments of a major medical center or university for evaluation.

TREATMENT ISSUES

Cleft Lip and Palate

The basic goals of CL/P treatment are quite straightforward: (1) to provide surgical closure of the lip while achieving the best possible appearance and function of the lip and nose, and (2) to close the cleft of the palate well enough and early enough to allow normal speech articulation and airflow while minimizing negative effects on tooth development and facial growth. However, in reality, achieving these simple goals can be complex. Parents of children with CL/P often feel confusion about the different treatment approaches that are advocated. With increased communication among parents because of Internet access and other sources of support organizations, the parents of an infant with CL/P may quickly realize that there are many different approaches to the treatment of their child's CL/P. They learn about feeding appliances, nasal molding appliances, active and passive presurgical appliances, early versus late bone grafting, and so on.

Parents may feel that they need to become “experts” at cleft surgery so that they are able to make informed choices. They worry that they will make the wrong choice and their child will suffer irreparable damage. Consequently, it is critical that parents be given the message that there are many valid and acceptable ways to address the surgical treatment of their child. Honest acknowledgment of the different approaches, including recognition that other approaches can be successful, while expressing confidence in the approach that your team uses, will reassure the family. Parents can be counseled that “We are confident in the approach we take in our team; however, other teams may take a different approach and still obtain very good surgical results. There are many paths that lead to a similar outcome.” Justification for your approach, with good outcome data, will ease parental anxiety and help them make an informed choice.

Craniofacial Conditions

The timing and frequency of surgical intervention is an important issue that affects surgical care across diagnostic categories. The surgical needs of children with CFCs are often very complex, and it is not uncommon for some children with craniofacial syndromes to have 20 to 30 surgeries during childhood. In these situations it is often difficult to balance the child's emotional needs with treatment objectives. Each time a child is hospitalized, regardless of the length of the hospitalization or seriousness of the procedure, the child and family must cope with a variety of stressors. The general rule is that the fewer surgeries a child must experience, the better for the emotional development of the child and family.

General Considerations

Children react in diverse ways to the recommendation for surgery. Even if the child desires improvement in appearance or functioning, the idea of blood tests, intravenous lines, and anesthesia is stressful and may add tension to the parent-child relationship. Surgery also interrupts a child's social life, including peer relationships, school activities, and athletics. For some parents a recommendation for surgery may bring back all the emotions associated with the child's birth, regenerating feelings of sadness, anxiety, and guilt. In addition to these stresses, parents must also cope with time off work, care of their other children, financial worries, insurance hassles, and the logistics of transportation and lodging during the child's hospital stay. Consequently, it is critical that families play an active role in determining the focus and timing of surgeries.

Surgeries for CL/P and CFC are generally recommended in stages across childhood. There are some surgeries that are time specific, such as a craniotomy, secondary management of the palate to improve speech, a bone graft timed to coincide with tooth development, or midface

advancement. However, many of the procedures, while clearly medically indicated, can be done when the child and parents are ready. Factors to be considered when discussing these types of time-elective surgeries include the child's age, temperament, psychological adjustment, motivation, and expectations, and the parents' ability to encourage and support their child.

There are individual differences in children that are developmental, physiologic, and temperamental.⁴¹ Both genes and environment influence these individual differences. Physiologically, some children may react to stress with a markedly increased heart rate or by vomiting, whereas other children will react to the same situation with less intensity. The intensity of the physiologic reaction has a biologic basis and is less under the control of the child. Children also differ on temperamental dimensions, such as vigor of motor activity, tolerance for frustration, quality of mood, attention span, and degree of inhibition. Both physiologic and temperamental characteristics have a biologic basis; in addition, the reactions of parents and other adults to the child's behavioral responses when under stress also contribute an environmental stimulus that may influence the child's long-term adjustment.³

Three broad personality types that have their basis in physiologic reactivity, temperament, and behavior and that have been reliably replicated for large groups of children have been identified by child development researchers.⁴² These personality types can be described as the *resilient*, *overcontrolled*, and *undercontrolled child*. The resilient child is defined as socially facile with adults and children, lacking in shyness, and possessing positive emotions. A child who fits the overcontrolled type personality is extremely shy, somewhat anxious, compliant, and dependent. At the other extreme, the undercontrolled child is uncooperative, more likely to be impulsive and non-compliant, displays predominantly negative emotions, and has social problems with peers. These personality types are present on a continuum, so these characteristics can be more or less defining for an individual child. The child's innate personality will influence the way he or she will behave, think, and feel in many situations and thus may shape the surgical experience.

Two- to Five-Year-Old Children

A younger child can successfully undergo a lip revision if the preschooler is calm and cooperative (characteristics of the resilient child) and the child's parents are able to provide emotional support and behavioral guidance. An energetic, active toddler or preschooler who is always on the go (even if otherwise similar in personality to the resilient child) is more likely to accidentally injure the surgical site through a fall or some other mishap during the recovery period and is probably not a good candidate for lip revision at a young age regardless of parenting skill.

A toddler or preschooler who is extremely shy and thus uncooperative during the clinic evaluations (overcontrolled) would probably also benefit from waiting until an older age for elective revisions. An overcontrolled preschooler is more likely to experience excessive anxiety and stress related to the various aspects of hospitalization and surgical care. Younger children do not have the cognitive abilities to actively participate in decision-making regarding a procedure, nor do they have the psychological skills to cope effectively with the stress of surgery, but they do understand that things are being done to them that they do not like. If the parents of a child who fits the description of the overcontrolled child are themselves anxious and dependent, they will have difficulty providing adequate support for their child. However, parents who are warm, encouraging, and patient with an anxious child may be able to provide an environment that will allow their child to successfully tolerate a surgery at a younger age. Thus individual decision-making is the key when working with a child who fits the overcontrolled personality type.

There are some preschoolers who are truly difficult to manage. These children may have been more irritable as infants, and may have a lower tolerance for stimulation, a high activity level, and intense reactions to new people and environments (the undercontrolled child). In general, an undercontrolled child is not a good candidate for elective surgery during the toddler or

preschool years. If a surgery must be performed on a young child who fits the undercontrolled personality type, both parents and hospital staff should be prepared for the possibility of strong reactions from the child. An undercontrolled child is more likely to pull out an IV line, injure a surgical site, or refuse to eat or drink after surgery. To counter these behaviors, presurgical planning and parent counseling may be indicated; however, even under the best of circumstances, it will be difficult to support an undercontrolled child through surgery at a young age.

If a toddler or preschooler is to undergo surgery, special support will be needed to make the surgical experience as stress free as possible. Parental involvement can be very beneficial, particularly when parents recognize the temperamental characteristics of their own child and are able to support the child appropriately when the child is stressed. Parents can be taught to distract their child in a positive way during potentially stressful procedures such as blood drawing or wound cleaning. Parents should be encouraged to cuddle and reassure their child through simple words that communicate confidence (such as “You’re doing well,” “Mommy’s here with you,” “Let’s read, sing, play,” and similar messages). Parents and professionals all need to recognize that young children express distress and fear through crying, resistance, and withdrawal. Hospitals are frightening places, and many children will be distressed during a hospitalization. Parents and surgeons can help children cope more effectively with the stress of a hospitalization if they acknowledge children’s fears as valid while simultaneously providing the emotional support they need. Young children especially benefit from opportunities to use play to express fears, and hospital-based therapists (such as pediatric psychologists or child-life therapists) can be consulted for help in this area.⁴³

School-Aged Child

A child’s entrance into elementary school is a major milestone. Families and surgeons share the goal of having a child look as normal as possible when that child begins formal education in the hope that a more normal appearance will increase the child’s positive social reception and minimize negative attention and teasing from peers. Certainly every child deserves to look as good as possible as they approach elementary school age. When a child enters first grade, they will find themselves rubbing shoulders with children of various ages, many of whom will be more aware of differences in appearance than the child’s preschool classmates have been. Consequently, the potential for teasing, bullying, and social exclusion becomes greater. However, teasing and negative social contacts are not just due to appearance issues. There have now been several studies that found children with CL/P to be more inhibited and more anxious, more socially isolated, and less friendly to peers than unaffected children.^{9,12,44} Murray et al⁹ reported that the primary correlate of these social difficulties at age 7 was the child’s ability to communicate effectively, including both articulation and the pragmatic aspects of communication (eye contact, tone, and volume of voice) and not the child’s appearance. Thus it is critical that improvement in appearance be presented as only one factor affecting social acceptance for the school-aged child.

Certainly we want children with facial differences to have the benefit of the best possible appearance; however, we do families a disservice if we encourage them to think that improved appearance will cure their child’s social difficulties or prevent peer teasing. Most children are teased at some time in childhood, and many children with facial differences do not experience teasing about their faces.¹⁶ Even children with facial differences who are being teased are not always being teased about their face. The causes of the social difficulties are multifaceted; therefore it may be better to present the timing of surgical revisions as occurring in the context of physical and psychological maturation rather than relate them to school entrance or prevention of social problems. Parents are rightly concerned about the possibility of teasing when appearance is deviant; however, whenever surgical revisions are discussed in response to concerns about teasing, members of the cleft-craniofacial team should take the time to talk about the prosocial skills that

children with appearance differences can use to minimize negative social interactions. For example, parents could be encouraged to consider how their child approaches peers and how the child responds to the social approaches of others. These discussions need to be framed so that the child is not “blamed” for teasing that occurs. The focus can be on broad factors that influence social acceptance for children, particularly social interaction skills that promote friendship, such as making eye contact, speaking with appropriate volume, greeting peers in a warm and gracious manner, responding appropriately to the initiation of peers, and knowing how to bring up topics of conversation that are of interest to peers.^{4,12,21} As previously mentioned, parents should also be encouraged to speak with school personnel to ensure that they have programs in place to prevent school bullying.¹⁷

Personality types continue to play a role as children enter elementary school. Professionals often wonder why a child with minimal scarring or facial deviance experiences excessive teasing, while another child whose scarring or deformity is much more severe is not reporting difficulties. The latter child most likely fits into the resilient personality type. That child is more likely to have a friendly approach to peers, to know how to share and give compliments, and to have a generally positive mood. When questioned about facial differences, a child with a resilient personality type is not likely to take offense, but will answer questions directly, with dignity and confidence. For children who fit the resilient personality type, surgical revisions tend to be viewed as a way to improve appearance, not as a way to make more friends or prevent teasing.

Children who are overcontrolled are most often viewed as “very well-behaved children” in school. They are quiet and may also be very conscientious. Unless a child who is overcontrolled also has learning disabilities, school achievement is likely to be good. However, children who are overcontrolled are also more likely to withdraw from social contacts. They may regularly choose to play alone, put their head down and refuse to speak when another child addresses them, and may display other signs of anxiety. This type of child is quite vulnerable to taunting from peers. Children who are overcontrolled may benefit from direct teaching of social skills to help them overcome the extreme shyness exhibited. Improving the appearance of a child’s face without also addressing the social inhibition issue is unlikely to affect the patterns of social interaction that have developed in the peer group.

A child who fits the undercontrolled personality type is at higher risk for global behavior disorders in general and externalizing disorders in particular during the school years.⁴² Even if the child does not have a specific learning disability, the undercontrolled child is more likely to have achievement problems that are related to behavioral issues (attention, motivation, and/or compliance) and is at greater risk for being openly rejected by peers because of the tendency toward aggression, difficulty learning to share or focus on another’s needs, and negative emotions. The presence of facial differences may become the focal point of “blame” for the child’s behavioral and social difficulties, and some families may hope that an improved appearance will have a positive effect on the child’s adjustment. Experience suggests that these families are generally disappointed unless direct action is also taken to address the underlying behavioral issues that are the primary source of the child’s problem. In this type of situation, a referral for a psychological consultation to determine an appropriate psychotherapeutic intervention is critical. Often the combination of psychological and surgical intervention can make a significant difference in the child’s overall adjustment.

Adolescents

Adolescence is a time of turbulence for many youths. Issues of identity, independence, and sociability are the major tasks of this age group. Relationships with adults and parents are redefined

as the adolescent seeks to establish a sense of self that is separate from the family of origin. The values of friends are compared with those of family members and sometimes chosen over those taught within the home. An individual's perception of personal success or failure may depend on his or her status in the social group.⁴⁵

Issues related to objective differences in facial appearance complicate the already complex tasks of adolescence for teens with CL/P or CFC. Teens must decide whether they will allow the presence of facial differences to interfere with social relationships, including dating relationships, during the high school years. Self-consciousness regarding appearance is normative during adolescence; however, when a teen has visible facial differences, the level of self-consciousness may intensify. Surgery may be desired to achieve the positive differences that are anticipated; in contrast, surgery may be feared because of the additional attention it will bring to the teen. Surgery may also interfere with activities that teens enjoy, such as sports, playing a horn or reed instrument, and summer jobs. Decisions regarding surgery create additional tension for teens. There may be disagreements between the teen, parent, and surgeon about surgical priorities or even whether additional surgery should be performed.

Few studies have objectively examined the rate of agreement between parents and adolescents regarding the importance of elective procedures to improve appearance. However, in a well-designed study, Turner et al⁴⁶ essentially found no agreement between parents and their 15-year-olds regarding satisfaction with cleft-related facial features. Based on paired comparison, parents expressed significantly greater overall satisfaction than the adolescents regarding the outcome in cleft-related areas. If parents and teens view treatment outcomes differently, who should control the decision-making process regarding additional elective treatment during adolescence?

Strategies are available to help parents and adolescents resolve differing views about the end-point of cleft-craniofacial care. The "Self-Understanding Model" provides a structure for helping families articulate differences of opinion regarding additional treatment.^{20,45} Using this model, both teens and parents are able to identify their own motivations for having or not having additional surgical treatment. Disagreements between parents and teens frequently occur because of misperceptions of motivation and false attribution of the meaning of additional surgery to the family. Open communication, encouraged and guided by members of the treatment team, can help families determine treatment priorities. This type of direct communication may help preserve the self-esteem of the adolescent and ensure that planned surgeries are being performed with the teen's cooperation. Research also suggests that active participation in decision-making by the teen results in increased satisfaction with the surgical outcome.⁴⁷

CONCLUSION

The treatment of CL/P and CFC extends over the infancy, childhood, and adolescence of individuals who are born with this condition. The children and their families contend with multiple surgeries throughout these years. Depending on the nature and severity of the condition, the families may also need to cope with ear infections, hearing loss, feeding tubes, speech therapy, learning disabilities, behavior problems, and various orthodontic treatments. The end result of these treatments and interventions should be a child, teen, and adult who is medically and psychologically healthy and an appropriately contributing member of society at each stage of development.³ The psychological support needed to enable a child to meet that goal should be provided by the family, surgeon, psychologist, school, and other members of the cleft treatment team. These adults should demonstrate a belief in the child's ability to cope with the challenges of medical treatment related to the CL/P or CFC and should focus the child on the efficacy he or she will gain from having had the experience of growing up with a CL/P or CFC.

KEY POINTS

- Medical professionals can influence the expectations of parents who have newborn infants with facial differences.
- Children should participate in the surgical decision-making process as appropriate to their developmental age.
- A young child's ability to cooperate with surgery is influenced by innate temperament, hospital environment and support, and the family's parenting skills.
- Developmental, physiologic, and temperamental differences in children affect their ability to cope with recommended medical treatments.
- Children with facial differences can be taught positive social interaction skills that encourage positive peer relationships and promote psychological adjustment.
- Children with CL/P and other CFCs are at higher risk for specific learning difficulties, which can be overlooked by teachers.
- There is growing evidence that learning disabilities in children with CL/P and other CFCs have a neurobiologic basis.

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18

Craniofacial Syndromes: Genetics, Embryology, and Clinical Relevance

Adel Y. Fattah

Understanding Genetics and the Basis of Development

The development of the craniofacial complex is fascinating for its convergence of multiple interacting systems in a small space. This chapter is divided into two sections: the first section will outline the basics of genetics for surgeons and introduce the principles of development pertinent to congenital malformations of the head and neck. The second section will provide an overview of craniofacial development by region, relating the developmental processes to the clinical conditions to foster a deeper understanding.

Molecular Biology's Influence on Gene Function

The power of molecular biology techniques and the science of experimental embryology unified to create the discipline of developmental biology. This technologic advancement has allowed scientists to alter specific genes, insert them into model organisms, and evaluate the changes they produce during embryogenesis. This has led to the publication of a significant number of papers describing anomalies in experimental animals that are analogous to human conditions. This in turn has helped direct clinical geneticists searching for candidate gene mutations when faced with an unknown craniofacial anomaly.

Box 18-1 Definitions

Congenital A feature existing at birth

Anomaly Any abnormality from the normal situation

Syndrome A collection of anomalies occurring in two or more embryologically distinct areas

Sequence A pattern of multiple anomalies presumed to be caused by a single factor

Malformation A morphologic defect resulting from a faulty developmental process

Disruption A morphologic defect caused by interference with a normal developmental process

Deformation A morphologic defect caused by a nondisruptive mechanical process

Dysplasia Abnormal development

Dysostosis Abnormal bone development

Familial Clustering of a particular feature in a family more than would be predicted by chance alone

Sporadic Any disorder occurring without a family history

Penetrance The proportion of individuals carrying a gene that displays an associated phenotype; if some do not exhibit signs and symptoms of the genetic disorder, it is said to have incomplete penetrance

Variable expressivity Variation of a phenotype within the same genetic disorder

Gene expression The manufacture of mRNA, which is then translated into the protein product by ribosomes

Molecular analysis is generally evaluated by the geneticist on the craniofacial team and is typically of limited interest to the surgeon, apart from confirming a diagnosis and knowing the impact that the diagnosis will have on the natural history of the child before and after surgery. However, genetic literacy will be important as we enter an age of molecular medicine, and doctors will require a working knowledge of the techniques and limitations of genetic testing. Furthermore, acquaintance with a few basic principles will allow a deeper understanding of the conditions we treat and empower surgeons to better conceptualize the pathologic basis of the anomaly. Some basic definitions are given in Box 18-1.

Gene Naming Conventions and Why the Names Are Often Strange

Many genes are named for what goes wrong when the gene does not work. Others are named for the protein for which they code, such as a fibroblast growth factor receptor. Many genes were initially identified in the fruit fly (*Drosophila*), with homologs identified in vertebrates: typically there is more than one version, which by evolution are co-opted for different functions. The fruit fly name for the gene is usually slightly modified for the vertebrate homolog. For example, a gene that causes the fruit fly larvae to have a prickly surface is called *hedgehog*. Three versions of hedgehog are found in vertebrates: *Sonic hedgehog* (SHH, a key patterning gene in multiple developmental processes), *Indian hedgehog* (involved in bone growth and fracture healing), and *Desert hedgehog* (loss of this leads to infertility). This history explains why many genes have seemingly unfathomable names. Such names are typically shortened to an initialism, with italicization and capitalization used to denote the species and whether one is referring to the DNA/RNA or the protein. Italicized gene names refer to the DNA or RNA; the nonitalicized version refers to the protein product. With some exceptions, using all capitals refers to the human species. If the first letter is capitalized, it refers to the mouse. If it is all lower case, then it refers to *Drosophila*, chick, frog (*Xenopus*), and zebrafish (*Danio*), which are the main model organisms. For example, SHH: human Sonic hedgehog protein; *Fgfr3*: mouse fibroblast growth factor receptor-3 DNA/RNA.

Box 18-2 Morphogenetic Mechanisms

- Cell division
- Cell differentiation
- Interaction of cells with extracellular matrix (for example, cell migration, branching of lung buds, invagination of otic placode)
- Induction: The effect of one tissue upon another signaling (for example, epithelial-mesenchymal interactions [tooth germ induction])
- Apoptosis

IMPORTANT CONCEPTS**Regional and Local Signaling Networks**

The morphology of the organism is created in a stepwise fashion after fertilization. The initial specifications of a body plan are given (for example, specifying cranial from caudal, left from right, dorsal from ventral). Thereafter, a cascade of signaling networks is set in motion to fashion the various parts of the organism; such regional signaling systems give way to local signals that are responsible for refining the pattern of the organism. For example, cell signaling positions the limb along the body axis, and then it patterns the limb, then the digits, and then the morphology of the skeletal elements within the digit.

As a consequence of this sequence, cells switch from a *pluripotent* (“stem cell–like”) situation to a commitment to the tissue to which the cells will contribute. How this is achieved is a function of their gene expression and is still only partially understood. Although signaling networks establish a body plan and the basic structures of the embryo, morphogenesis ultimately creates the form and shape of the organism and is distinct from cellular differentiation. Morphogenesis has a physical and mechanical basis, and current concepts in development are founded on the principle that differential gene expression leads to cellular interactions that create differences in morphology. Some mechanisms responsible for shaping the embryo are listed in Box 18-2.

Genes Make Proteins—Not Body Parts or Syndromes

Genotype refers to the genetic composition of an organism, whereas *phenotype* refers to the observable characteristics, which include morphology (that is, clinical signs), physiologic changes, and even behavior. The phenotype is in part dependent on the genotype, but also on environmental factors; as a consequence, the same genotype may result in different phenotypes (such as in identical twins raised in different environments).

Genes are transcribed into messenger RNA (*mRNA*), which is translated by ribosomes into amino acid chains that are assembled into proteins. The same genes and signaling pathways are used in many seemingly unrelated processes; for example, *SHH* is involved in many processes, including specifying the anteroposterior (radial-ulnar) axis of the limb, neural induction, and cholesterol metabolism. Therefore one of the most important concepts to grasp is what a gene does; a gene does not “make” an organ. If we remove *Pax6* from the genome, the eyes do not form.¹ However, *Pax6* does not code for an eye; it makes a transcriptional regulator that activates and represses a number of genes downstream that set in motion signaling processes and cellular interactions that culminate in eye formation.

Box 18-3 Nomenclature for Chromosomal Abnormalities (Paris Nomenclature)

p	Short arm (petit)
q	Long arm
13q32	Long arm of chromosome 13 (32 refers to the region of the chromosome)
del, D or Δ 22q11	Deletion of region 11 on the long arm of chromosome 22
t(3;13)	Translocation from chromosomes 3 to 13
t(3;13)(p21;q3)	Above translocation with the breakpoints specified

There Is No Gene for Apert Syndrome

By extension, there is no such thing as a gene for a specific syndrome, such as a gene for Apert syndrome. The gene mutated in Apert syndrome is a receptor for fibroblast growth factor (*FGFR2*) that transduces the FGF signal into the cell and initiates a cascade of intracellular processes. In Apert syndrome, this mutation accelerates endochondral ossification.² However, a different mutation in the same gene leads to reduced chondrocyte proliferation and a distinct syndrome called *Crouzon syndrome*.³ Environmental factors (for example, raised intracranial pressure) then act on this genotype to create the phenotype that is evaluated in the clinic.

The point is that genes do not code for syndromes; a syndrome refers to the constellation of features seen clinically. This apparent conflict arises from physicians and scientists trying to classify things from opposing perspectives. To a physician, a syndrome is a collection of clinical signs that may be caused by multiple different gene mutations; to a scientist, mutations in a gene may cause multiple different syndromes. At this point in our knowledge, there is no unifying classification; therefore syndromes and gene mutations will sometimes coincide, but sometimes they will not. It has been suggested that most patients can be assigned three diagnostic terms: phenotypic, pathologic, and genetic.⁴ For example, *plagiocephaly* is a phenotypic description, *unicoronal synostosis* is a pathologic diagnosis, and *FGFR3*^{P250R*} (Box 18-3) is a genetic description of the same condition.

Multiple Genes Have Overlapping Functions: Redundancy

Many important components in signaling, metabolic, and developmental pathways have multiple similar genes that appear to be functionally redundant. This is thought to be a consequence of genes being duplicated over evolutionary time. When one gene is inactivated, another is able to compensate as a result of the overlapping function. In humans, *ALX3* loss-of-function causes a midline facial anomaly called *frontorhiny*.⁵ By contrast, *Alx3* knockout mice have no anomalies, yet a double knockout of *Alx3* and *Alx4* creates a “split-face” phenotype. This example demonstrates functional redundancy (*Alx4* can compensate for the loss of *Alx3*), and furthermore, that the phenotype may differ between species and genetic backgrounds, as discussed later. Why we should have two genes when one will suffice has been a topic of debate. It has been suggested that this is a “belt and braces” fail-safe mechanism, or a process that modulates cell signaling.⁶

*The *P250R* informs the reader of where the mutation is. In this instance, a **P**roline is replaced with an **a**Rginine residue at the 250th position in the amino acid chain. Similar notation can be used for the DNA sequence; in this case, it is c.749C>G.

Relationship Between Genes and the Environment

Genes are not independent of their environment; some genetic effects are seen under certain environmental conditions, or the response to an environment may depend on the genotype. Genes are influenced by their environment in a number of ways; for example, ionizing radiation exerts its effect by directly breaking one or both strands of a DNA molecule, creating *de novo* mutations. By contrast, *epigenetic factors* are those that influence gene expression without altering the DNA. Epigenetic factors may be exogenous compounds or even chronic disease states that lead the body to produce certain factors that influence gene expression. How epigenetic factors affect gene regulation is an area of active research, but we know that they may affect any stage of the DNA-to-protein process and are a source of variation between normal individuals.

It is easy to forget that genes act within the environment of the cell, surrounded by a multitude of other gene products that can modify their behavior (the genetic background). Transcription factors are proteins that bind to the DNA and read the code, expressing it as an mRNA that will go on to make a protein. These factors may be sensitive to chemical signals that may be released in response to external stimuli, such as hormones and neurotransmitters that may alter gene expression. Such agents that disrupt normal development are called *teratogens*. Retinoic acid (vitamin A; Accutane) is involved in normal development, but high exogenous concentrations will cause a very specific pattern of anomalies in the fetus, dramatically reducing the first and second branchial arches. It appears to do this through its effect on *Hox* genes, limiting cranial neural crest proliferation and migration.^{7,8}

COMMON EXPERIMENTAL TECHNIQUES REFERRED TO IN THE LITERATURE

Understanding the function of a gene *in vivo* may be studied in essentially three ways: increasing the expression of a gene, reducing or removing its expression, or expressing the gene in a different place or time from the normal situation. In these instances, the sequence of the gene must be known before it can be manipulated and its function studied, an approach called *reverse genetics*.

Knockout and Knockin

Conventional (first-generation) *knockout mice* are created by a process whereby a gene of interest is replaced with nonfunctioning DNA. All cells in the embryo possess this defect, so if the gene in question is important for early development, embryonic lethality precludes study at later stages. By contrast, *conditional knockout mice* allow investigators to activate and deactivate genes in specific tissues or at specific times; this is called *targeted mutagenesis*. Similarly, *knockin* of gene function allows one to introduce a gene where or when it is not normally expressed and evaluate the outcome on development; this has allowed more focused studies of gene function.

Knockdown

Knockdown refers to a transient inactivation of gene function, which can be achieved by a variety of processes. A short sequence of nucleic acid (oligonucleotide; “oligo”) can bind to the gene and block transcription, bind to mRNA to degrade it (small interfering RNA [siRNA]), or block its translation into protein. This method allows one to switch genes off at specific times to elucidate function during specific developmental processes and may be used *in vitro* as well as in a number of animal models, especially zebrafish.

Gene Overexpression

Transgenic technology in species other than mice is limited, so overexpression of a gene has been produced by numerous other methods, including *retroviral overexpression*, which has been superseded by *electroporation*.⁹ Use of animal models such as the chick and zebrafish allows scientists to surgically manipulate the embryo as well as control gene expression, which is a powerful combination of techniques.

CLINICAL GENETIC TESTING: WHICH TEST FOR WHAT PROBLEM?

We will outline the basics of testing to arm the surgeon with sufficient information to converse with clinical geneticists and prepare for the new technologies that are on the horizon.

Genetic testing looks for differences between the genome of an affected individual and that of the normal population. These differences may be microscopically visible (for example, trisomy or large deletions), or they may be variations in the amount of a region of the genome that is not visible: *copy number variation* (CNV). Most humans have approximately 100 CNVs in their genome, and they are largely innocuous. *Single nucleotide polymorphisms* (SNPs) are changes to a single base of the DNA; every individual has about 3 million SNPs. Some are specific mutations that cause a genetic disorder, and some may predispose to disease, but most are benign. The complexity of genetic testing arises from differentiating normal genetic variation from disease-causing mutations.

Three general categories of testing exist:

1. *Cytogenetic*: Looks for chromosomal abnormalities
2. *Biochemical*: Analyzes proteins typically for inborn errors of metabolism
3. *Molecular*: Examines DNA sequences

Cytogenetic Testing: Karyotyping

Cytogenetic tests evaluate whole chromosomes for changes in number and structure and is the first tier of testing in a number of clinical situations. Perhaps the oldest technique still widely used, *karyotyping*, is the examination of chromosomes under a light microscope. The cells are lysed and fixed on a microscope slide and labeled with Giemsa stain to create the banding (G-banding) pattern that is characteristic of each chromosome, permitting identification. There is a standardized numeric system for these bands, and by looking at the banding pattern and comparing that with a normal karyotype, absence, duplication, and translocation of large portions of the genome (whole chromosomes, such as trisomy 21 or parts thereof) can be detected. These chunks of the genome are large and may span many specific genes as well as the intervening portions (*introns*).

Molecular Testing

In Situ Hybridization

Hybridization is the annealing (joining) of two complementary copies of DNA and the basis of virtually all molecular techniques. If one of these is labeled, for example, with fluorescence, this is called a *probe*, and one may visualize where these sequences are in tissues (in situ). *In situ hybridization* (ISH) is used to look at the presence of specific DNA or RNA sequences in different situations. To use this method, one needs to know the base sequence of what is being sought to synthesize the probe^{10,11} (Fig. 18-1).

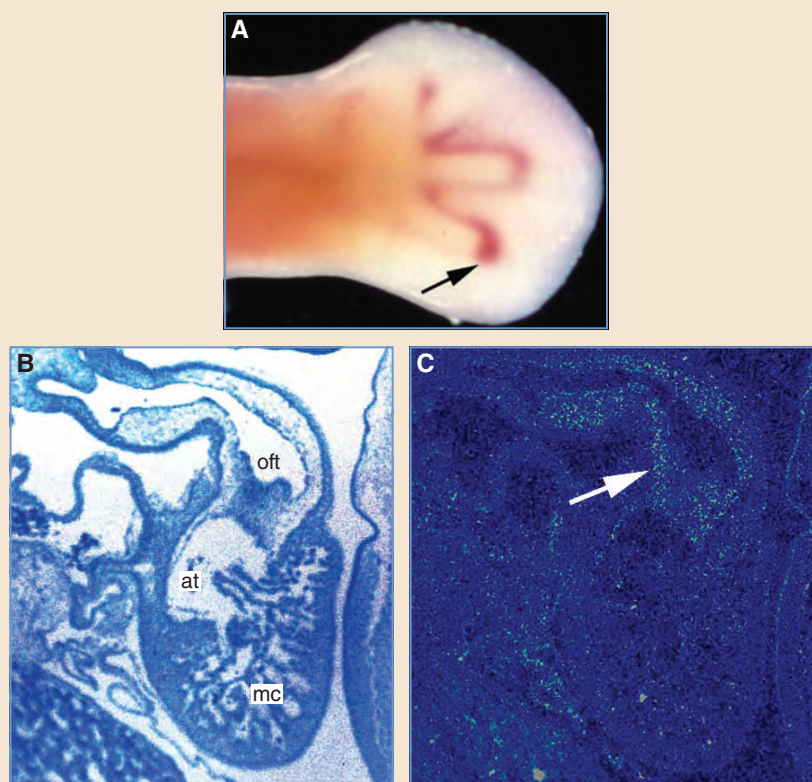


Fig. 18-1 In situ hybridization. **A**, Wholemount in situ hybridization of the forelimb with a probe to *Gdf3*¹ labeling the perichondrium and interphalangeal joints (*black arrow*). **B**, Light field view of embryonic heart. **C**, Radiolabeled in situ hybridization of the heart with probe for *frzbl*² labeling the neural crest derived cardiac outflow tract (*white arrow*). (*at*, Atrium; *mc*, myocardium; *oft*, outflow tract.)

Fluorescent In Situ Hybridization

Fluorescent in situ hybridization (FISH) is the most common form of ISH in clinical practice, whereby fluorescent probes are hybridized to a karyotype specimen. FISH is relatively sensitive, specific, and rapid. The label is observed under a fluorescent microscope and pinpoints where on the chromosome the complementary DNA sequence in question may be found. This can then be reported by standard cytogenetic nomenclature (for example, $\Delta 1q36$; Table 18-1). FISH cannot serve as a screening test; the clinician must be looking for a specific known genetic anomaly that can be confirmed or refuted by FISH. Therefore FISH is used to confirm a suspected clinical diagnosis, such as 22q deletion sequence, by the absence of fluorescence (it is deleted).

Comparative Genomic Hybridization or Chromosomal Microarray Analysis

Comparative genomic hybridization (CGH) or *chromosomal microarray analysis* (CMA) is another molecular method for analyzing gains and losses of DNA that are not detectable with routine karyotyping or FISH. The chromosomes are fragmented and arranged in a grid pattern so that each fragment can be identified; the relative degree of fluorescence relative to control DNA is analyzed. Differences in this are regions of genomic imbalance; it does not detect changes such

Table 18-1 Amino Acid Nomenclature

Symbol	Amino Acid	Symbol	Amino Acid
A	Alanine	L	Leucine
R	ARginine	K	Lysine
N	AsparagiNe (Asn)	M	Methionine
D	aspartic aciD (Asp)	F	Fenylalanine
C	Cysteine	P	Proline
E	Glutamic Acid (Glu)	S	Serine
Q	Qlutamine (Gln)	T	Threonine
G	Glycine	W	Tryptophan
H	Histidine	Y	TYrosine
I	Isoleucine	V	Valine

as translocations (where the DNA is simply moved to elsewhere in the genome), which do not alter the relative fluorescence. Arrays can be high or low resolution and be targeted to specific portions of the genome. Essentially, it is a similar principle to a G-band karyotype where one is looking for missing or additional chunks of DNA. The difference is that the resolution of CGH allows it to detect much smaller differences.

DNA Microarray

DNA microarray is “gene chip” analysis that involves a glass microscope slide to which the gene sequences of interest are bonded to act as probes (for example, candidate genes). To this the fluorescently labeled patient sample containing hundreds or thousands of genes is applied and will hybridize to the complementary DNA. This is called *interrogation*, and a computer is used to detect the areas in which patient and control DNA do not hybridize to the same degree. Efforts are being made to develop phenotype-specific panels so that many of the known mutations that produce a specific clinical picture (for example, syndromic craniosynostosis) can be interrogated at one sitting.

DNA Sequencing

If an array does not detect a disorder, then gene sequencing can be used in which the base sequence of the gene of interest is read and errors determined. This is the most time- and resource-consuming form of genetic testing. For about 25 years, Sanger (chain termination) sequencing¹² has been the technique used, but this has been superseded by next generation (NextGen) methods that are automated large-scale genome analyses. Such high-throughput genome sequencing technologies have been used as a research tool but are being introduced into clinics as the costs fall. *Whole genome sequencing* (WGS) sequences the complete genome DNA. By contrast, extracting and sequencing only the exons (the portions that are transcribed to be made into proteins) has proved to be fast and accurate; this is called *whole exome sequencing* (WES). But as the introns are no longer considered “junk DNA” and the costs continue to fall, WGS is likely to displace WES in the long term. To interpret these data efficiently, one needs to sequence the affected individual and both unaffected parents as a trio to quickly discard benign sequence variations (CNVs and SNPs). One of the greatest challenges of NextGen technology will be the bioinformatics required to store and analyze so much data. One caveat is that most SNP-based diagnostics are

probabilistic (based on a computerized statistical analysis of chemical reactions) rather than deterministic, such as the microscopic observation of a gene duplication. Nonetheless, we are less than a generation away from an individual's whole genome sequence being entered into his or her electronic patient record.¹³ Once this information is available, genetic "testing" will simply be a matter of comparing the patient's recorded gene sequence to control data.

CREATING A BABY: SPECIFYING A BODY PLAN AND EARLY CRANIOFACIAL DEVELOPMENT

Fertilization sparks a series of events, creating a complete human body plan within 8 weeks. To achieve this, a single cell needs to be instructed to divide and assemble in a coordinated manner. By the third week, gastrulation occurs whereby a flat three-layered sheet of the embryo is formed comprising ectoderm, mesoderm, and endoderm. By this process, rostrocaudal,* dorsoventral, and left-right axes are established, giving cells "positional information." On completion of gastrulation, a notochord (primitive vertebral axis) is formed that signals to the ectoderm to induce the neural plate, which gives rise to all neural tissue (Fig. 18-2). This flat plate rolls up to form the neural tube, which develops into the central nervous system and from which the neural crest arises and migrates, contributing to numerous key structures in the head and neck. Either side

*The rostrocaudal axis is that which runs nose to tail in all vertebrates. This avoids confusion with anteroposterior in humans (erect in the anatomic position) and quadrupedal species and thus refers to their craniocaudal axis.

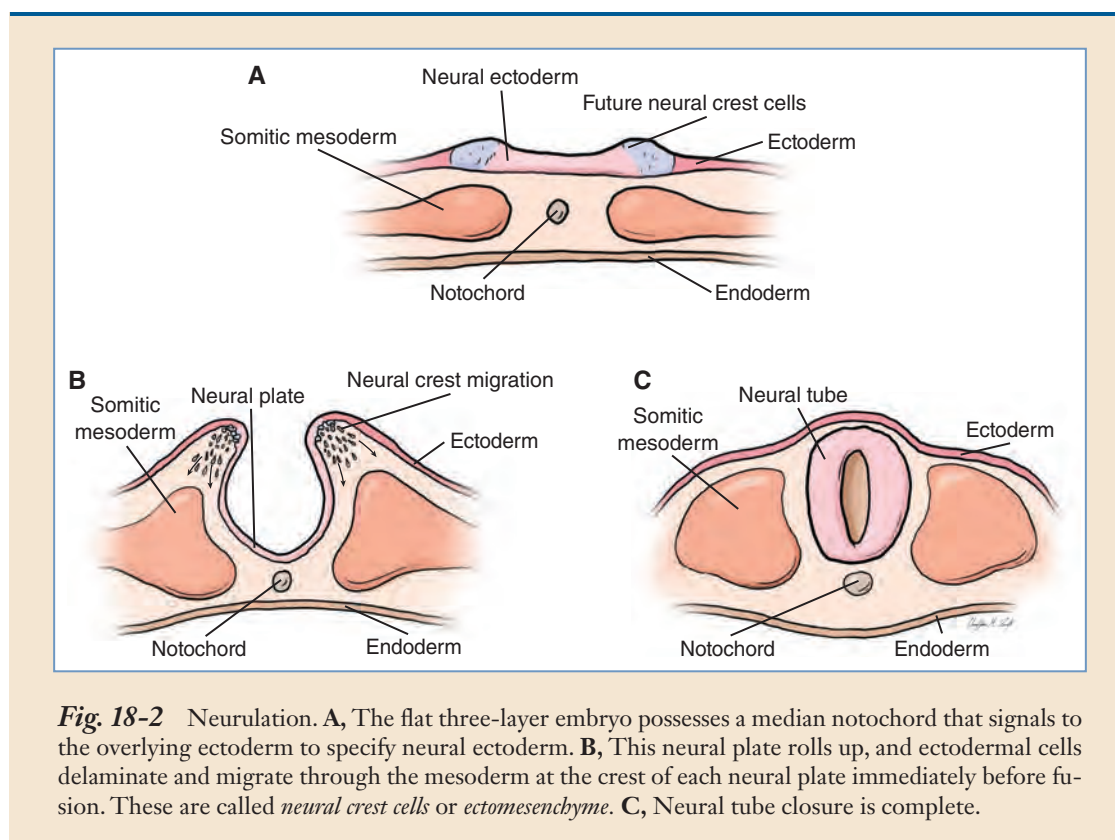


Fig. 18-2 Neurulation. **A**, The flat three-layer embryo possesses a median notochord that signals to the overlying ectoderm to specify neural ectoderm. **B**, This neural plate rolls up, and ectodermal cells delaminate and migrate through the mesoderm at the crest of each neural plate immediately before fusion. These are called *neural crest cells* or *ectomesenchyme*. **C**, Neural tube closure is complete.

of the neural tube lies the paraxial mesoderm that is segmented in register with the neuraxis. This somitomeric mesoderm migrates and gives rise to the musculature of the craniofacial region, angioblasts, and contributes to the skull base and calvarium.¹⁴

Neurulation and the Rostrocaudal Axis

Neurulation is the process by which the neural plate rolls up to form the neural tube. During this process, the neural crest is formed (see section below) and eventually a clear craniocaudal difference visibly emerges. The rostral neural tube expands and becomes subdivided in early development into the forebrain (*prosencephalon*), midbrain (*mesencephalon*), and hindbrain (*rhombencephalon*). By the fifth week of gestation, this simple three-segment brain further develops, with the prosencephalon dividing into the rostral *telencephalon*, which will form the cerebral hemispheres, and the caudal *diencephalon*, which is recognizable by the optic vesicles extending from either side of it. The mesencephalon remains undivided, whereas the rhombencephalon (comprising segments called *rhombomeres*) can be morphologically divided into the *metencephalon* (future cerebellum and pons) and more caudal *myelencephalon*^{3,15,16} (medulla) (Fig. 18-3).

Hox Genes and Rostrocaudal Specification

In the trunk, each of the thoracic, lumbar, and sacral levels represents a unit of metameric segmentation, whereas in the cranial region, such segmentation is not morphologically obvious. The debate regarding the segmentation of the head has a long history. The first experimental evidence that segmentation exists in the cranial region was presented by Lumsden and Keynes. They demonstrated that the pattern of rhombomeres corresponded to the arrangement of the roots of the cranial nerves.¹⁷ This subdivision was subsequently supported at the molecular level by the expression of *Hox* genes, a family of homeodomain-containing genes that encode protein regulators of gene transcription.

Four (paralogous) groups (A through D) of sequentially numbered *Hox* genes (1 through 13) occur in humans. They are unique in that they are expressed in combinations along the longitudinal axis of the neural tube, each having a different cephalic boundary of expression¹⁸ (see Fig. 18-3). The different combinations of *Hox* genes at a specific point along the craniocaudal axis confer specific information relevant to that position.¹⁹ For example, retinoic acid is known to move the boundary of expression of *Hoxb-1* cephalically along the rhombencephalon. This changes the regional character of rhombomeres so that instead of giving rise to the trigeminal nerve, they now produce a duplicate facial nerve.²⁰ These homeotic genes are thus regulators of developmental patterning; by encoding transcription factors that influence batteries of other genes, they can be broadly considered master genes for regional specification.

The Neural Crest

The *neural crest*, which carries positional information from the brain into the face, is a key component of the head and neck, forming much of the connective tissues and peripheral nervous system of the head. This includes bones, cartilages, cranial ganglia, odontoblasts, corneal stromal fibroblasts, muscle fascia, vascular smooth muscle, dermis, and melanocytes. The neural crest arises as a distinct cell population at the border between neural and nonneural ectoderm. As the neural folds rise up to reach each other, but before their fusion to form the neural tube, the cells at the crest of this fold begin to delaminate, beginning at the level of the mesencephalon at 3½ weeks' gestation. These neural crest cells pass through interruptions in the basement mem-

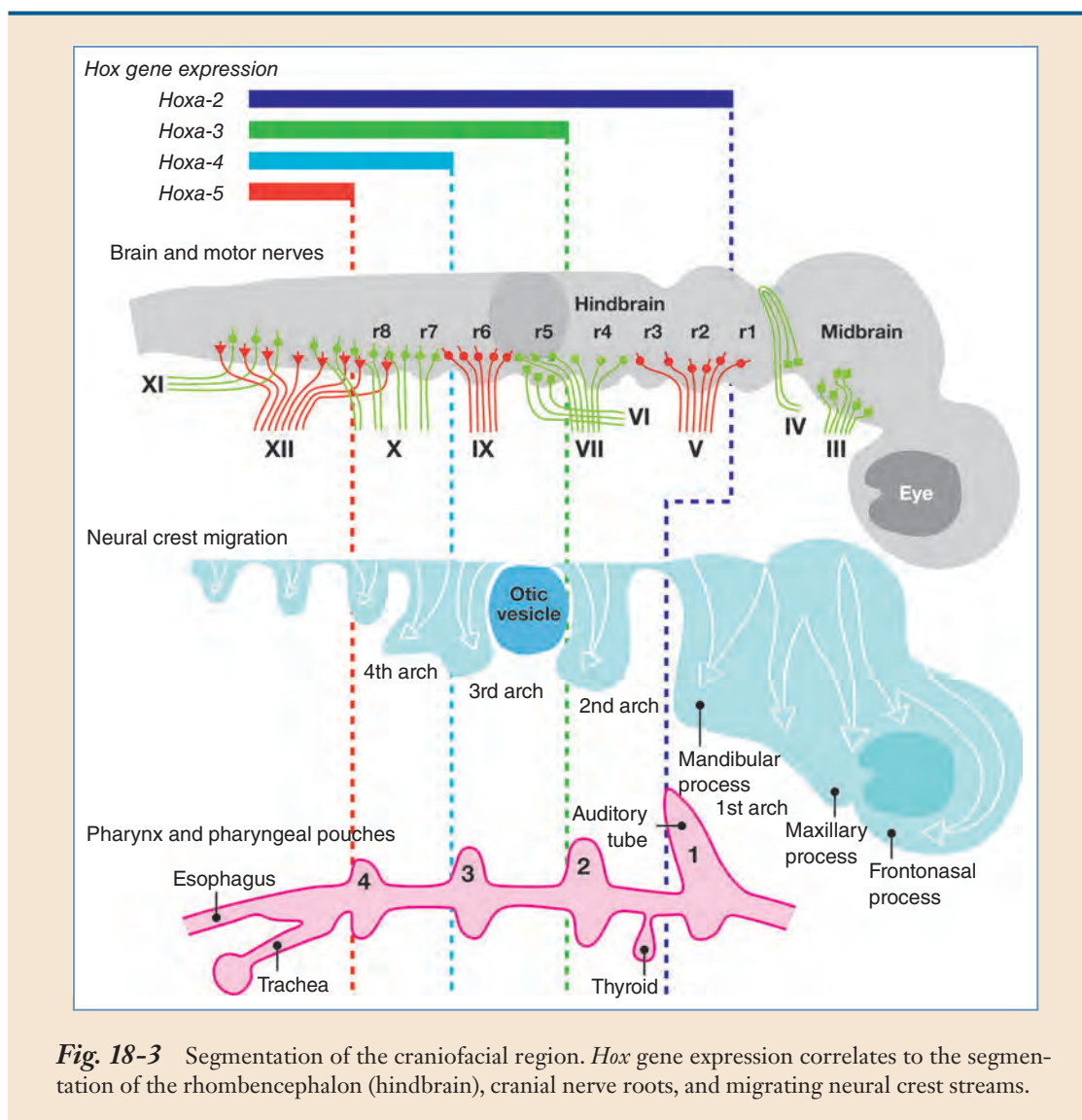


Fig. 18-3 Segmentation of the craniofacial region. *Hox* gene expression correlates to the segmentation of the rhombencephalon (hindbrain), cranial nerve roots, and migrating neural crest streams.

brane to enter the mesodermal layer, at which point they migrate to form a substantial proportion of the facial mesenchyme. Studies reveal that migration follows specific routes with a craniocaudal correlation between the origin and destination of the neural crest. Ephrins and their receptors are mediators of cell contact-dependent interaction and have an important role in creating defined streams of cell migration.²¹ It may be considered that three major migratory pathways are taken by these cells^{22,23}:

1. The rostralmost stream migrates around the optic vesicle and contributes to the fronto-nasal process and the maxillary and mandibular processes of the face.
2. The otic vesicle divides the remaining cranial neural crest into the preotic crest, which migrates into the hyoid arch and contributes to the facial nerve ganglia.
3. The postotic crest that fills the third and fourth branchial arches contributes to glosso-pharyngeal and vagal nerve ganglia and the heart (cardiac neural crest), where it surrounds the truncus arteriosus (see Fig. 18-1).

These migrating neural crest cells express a combination of *Hox* genes appropriate to their origin from the rostrocaudal axis of the neural tube and carry this information into the branchial arches. The cells proliferate and signaling cues guide migration to their destination, where they are influenced by local signals; a failure of these processes to occur in an appropriate spatiotemporal sequence gives rise to a wide range of congenital abnormalities.²⁴ Therefore the neural crest can simplistically be considered to be carrying genetic information into the face and branchial arches to allow regional programs of development to be initiated.

Neurocristopathies and CHARGE Syndrome

Neurocristopathies are disorders of neural crest development and are broadly divided into defects of migration and morphogenesis (such as CHARGE syndrome) and tumors arising from neural crest tissues (such as neurofibromatosis). Given the major contribution of the neural crest to the craniofacial complex, a number of craniofacial malformations may be called *neurocristopathies*; these include hemifacial microsomia, 22q deletion syndrome (22q DS), DiGeorge syndrome, and Treacher Collins syndrome (discussed later in this chapter under Branchial Arch Syndromes).

CHARGE syndrome demonstrates the breadth of effects caused by these disorders; the acronym summarizes the key features: coloboma, heart defects, choanal atresia, retardation of growth and development, genital anomalies, and ear anomalies. Major and minor diagnostic criteria exist²⁵ (Fig. 18-4). Most cases are sporadic and 90% are associated with haploinsufficiency of *CHD7*, a transcription factor that interacts with genes essential for neural crest migration.²⁵ The other 10% of cases are thought to be mutations in other genes and the same features can be produced by teratogens such as retinoic acid; therefore, CHARGE is still a clinical diagnosis rather than a genetic diagnosis.²⁵

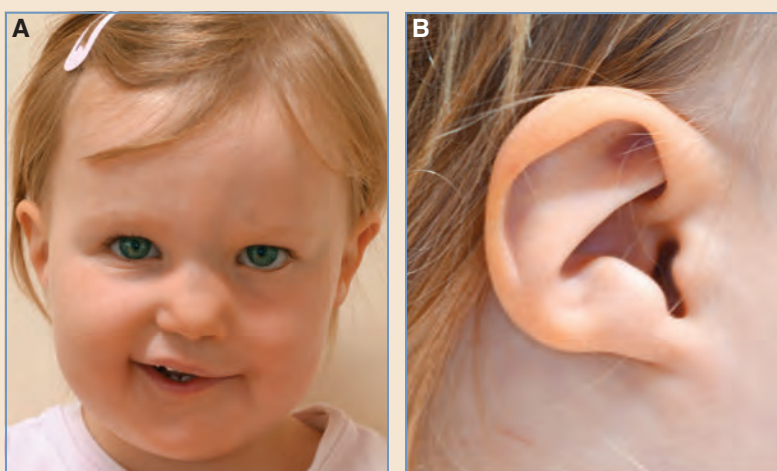


Fig. 18-4 A child with CHARGE syndrome. **A**, Typical facies with prominent forehead, broad nasal bridge, and left facial palsy. Note the similarity to the facies of patients with 22q DS. **B**, Characteristic triangular concha.

CHARGE patients are most likely to present to the ENT surgeon with choanal atresia or to the craniofacial team with cranial nerve dysfunction (for example, facial palsy), external ear anomalies (triangular concha), or cleft lip and palate (CL/P). Each of these issues is managed as in other individuals, but multidisciplinary coordination of care improves the family and patient experience. Feeding difficulties may be caused by lower cranial nerve palsies and may necessitate tube feeding. In addition to sensorineural hearing loss, semicircular canal anomalies may present as a lack of motor coordination, with delayed milestones but rarely dizziness. Most of these children are intellectually normal or near normal. Echocardiography and ultrasonography of the kidneys should be performed in all patients to screen for anomalies where early intervention may be important. Educational psychology is important in managing the developmental and behavioral aspects of this disorder.

CONCLUSION

As a consequence of neurulation, neural crest migration, and movements of the somitomeric mesoderm, the early cranial region comes to be dominated by the primitive brain dorsal to the notochord with the pharyngeal region below. At approximately the fourth gestational week, the head fold develops, and subsequently a number of bulges develop around the primitive mouth (stomodeum) that comprise the facial processes and pharyngeal arches. This primitive segmental arrangement undergoes great morphologic change to become the recognizable structures of the head and neck. Subsequent craniofacial development can be subdivided into development of the brain and midline facial development, fusion of the facial processes and palate, development of the pharyngeal arches, cranial vault development, and development of the cranial base. Each will be described in turn in the next section, along with its clinical correlates.

Concepts for Understanding Congenital Facial Differences

The basic structure of the face is formed between the fourth and eighth weeks of gestation, but facial development continues into puberty, once dentoskeletal maturation is complete. Toward the end of the fourth week, five processes appear at the base of the prosencephalic vesicle that will eventually unite to form the face (Fig. 18-5). A midline process called the frontonasal process and the lateral paired maxillary and mandibular processes (derived from the first pharyngeal arch) surround the stomodeum to unite to form the facial structures. By the fifth week, the frontonasal process is flanked on either side by the nasal placodes and will form the intermaxillary segment, a precursor of the philtrum of the upper lip, the premaxillary part of the upper jaw, and the primary palate. By the end of the sixth week, the frontonasal process engulfs the nasal placodes, creating a hooded appearance as each placode sinks into the underlying tissues to create the olfactory pit. The frontonasal process now consists of both lateral and medial nasal processes, and the olfactory pit communicates with the buccal cavity by the nasobuccal channel. Meanwhile, the maxillary process meets the lateral nasal process and fuses with it, and by the seventh week, it meets the medial nasal process and its fellow of the opposite side. Thus the union of the upper maxillary processes is completed.

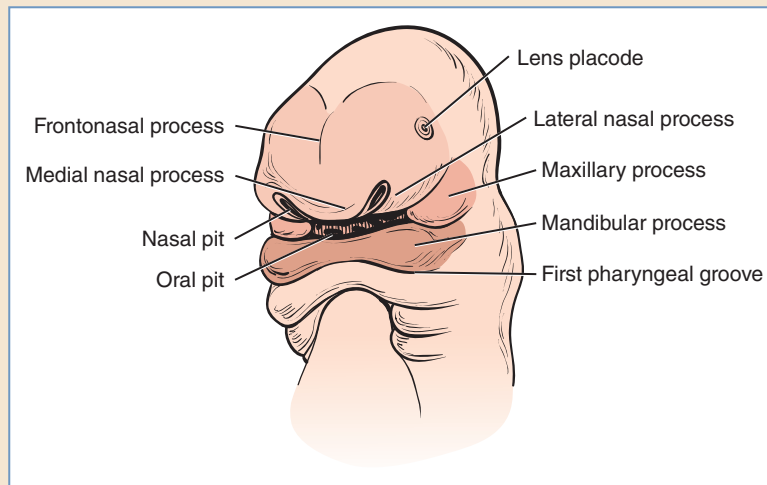


Fig. 18-5 The facial processes.

Classification Based on Developmental Processes

Various classifications of craniofacial conditions are often complex and confusing and are rarely widely adopted. My former career as a developmental biologist has led to a personal working classification based on the mechanisms of craniofacial development. This allows the developmental mechanisms, genes, and syndromes to be related to each other and provides a framework on which to associate new clinical, genetic, and scientific information.

MIDLINE FACIAL DEVELOPMENT

The development of the frontonasal process is a key component of normal facial morphology. However, signals to pattern the midline of the face arise at gastrulation, when an area rostral to the notochord called the *prechordal plate* is formed. Experimental evidence demonstrates that midline facial development involves a stepwise mechanism, relaying signals from the prechordal plate to the ventral forebrain (prosencephalon), onto the migrating neural crest, and then the facial ectoderm, which finally establishes signaling centers around the olfactory pit that influence the morphology of the developing face (Fig. 18-6). Experimental manipulation (surgical or genetic) of this process can produce midline facial excess or deficiency, mirroring the clinical spectrum of orbital hypertelorism and holoprosencephaly (HPE), respectively; the earlier in development this is performed, the more severe the phenotype. This stepwise mechanism intimately links the development of the brain with the facial midline.

The formation of the prechordal plate requires the signaling of Nodal in concert with TDGF1 through the TGF β signaling pathway, together with a repression of bone morphogenetic protein (BMP) activity.²⁶⁻²⁸ Once established, the prechordal plate signals to the overlying ventral prosencephalon by synthesis of the secreted signal encoded by SHH. The SHH signal activates the homeobox gene *Six3* in the ventral forebrain, establishing a feedback loop that maintains SHH expression.²⁹ This forebrain SHH signal is communicated through the migrating neural crest to the facial ectoderm, where a new SHH signaling region is initiated. Inhibition of

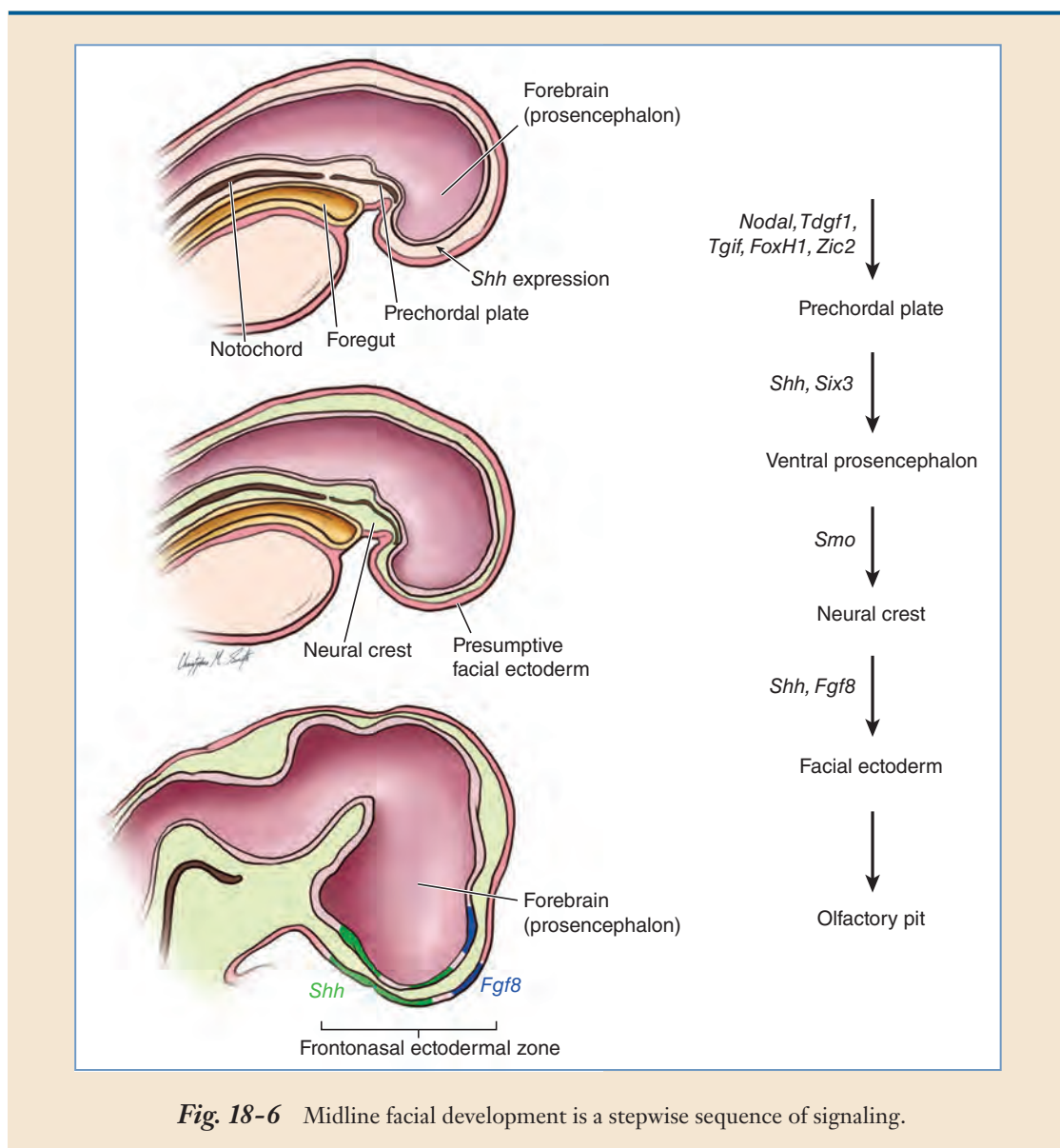


Fig. 18-6 Midline facial development is a stepwise sequence of signaling.

this SHH activity or surgical removal of the neural crest resulted in a failure of the frontonasal process to develop normally and anomalies resembling HPE.³⁰

Ectodermal expression of SHH and adjacent *Fgf8* create a signaling region called the *frontonasal ectodermal zone*, which appears to coordinate patterned outgrowth of the frontonasal process.³¹ Excision of this region leads to hypoplasia of the frontonasal process and a facial cleft phenotype. By contrast, application of SHH protein or overexpression of SHH lead to orbital hypertelorism and duplicated midline structures.³² Once the frontonasal process has formed, FGF signaling around the nasal pits appears to be vital to morphogenesis. Inhibiting their activity leads to apoptosis and midfacial hypoplasia, whereas application of FGF2 leads to increased cell proliferation and an enlarged frontonasal process.³³

Median Facial Dysgenesis With Deficiency: The Holoprosencephaly Spectrum

Midline defects of the craniofacial complex have been described by Tessier³⁴ as 0-14 clefts in which he includes conditions with deficiency and excess. Deficiency includes orbital hypotelorism, arhinencephaly, and absent premaxilla and prolabium together with various cleft lip configurations. Such features are part of the *holoprosencephaly* (HPE) spectrum. HPE is a defect of the forebrain in which there is a failure of cleavage of the prosencephalon into the paired telecephalic vesicles that form the cerebral hemispheres. It is etiologically heterogeneous and a feature of a number of syndromes. It is classified neuroanatomically as *alobar* (a single monoventricular cerebrum), *semilobar* (rudimentary cleavage of the cerebrum), and *lobar*, where there are two cerebral hemispheres, but callosal hypoplasia or agenesis may occur. Pituitary dysfunction, particularly diabetes insipidus, is common and should be actively managed. Traditionally, it is said that the “face dictates the brain,” in which the severity of the facial deformity is correlated to underlying cerebral pathology, but this is not the situation in approximately 20% of cases.^{35,36}

Cyclopia is the most severe facial phenotype in which there is a median eye, proboscis, and the nose is absent. *Ethmocephaly* comprises arhinia, proboscis, and paired but hypoteloric eyes. *Cebocephaly* is the presence of a single nostril pit, and finally, there is median cleft lip, which is associated with absence or hypoplasia of the primary palate. Not all median cleft lips are associated with brain anomalies, and individuals with normal intelligence and head circumference are said to have *binderoid clefts* and are not considered holoprosencephalic.³⁷ Microforms exist, including a single maxillary incisor and the absence of septal cartilage, labial frenulum, and philtrum. However, diagnosis at this subtle end of the spectrum becomes problematic, because many of the features may occur without central nervous system anomalies, and the diagnosis of HPE may depend more on the context in which the abnormality is diagnosed.^{36,38} Although many clinical cases can be categorized accordingly, the clinical presentation of HPE is in fact a spectrum ranging across this classification.^{36,39}

Gene mutations have been identified in roughly 20% of cases with variable expressivity and penetrance. Many of these genes are components of the signaling pathways outlined above: *NODAL*,⁴⁰ *TDGF1*,⁴¹ *SIX3*,⁴² *SHH*, and its receptor *PATCHED1*,⁴³ as well as its downstream target *GLI2*.^{44,45} In very general terms, mutations that reduce the SHH signal lead to deficiency and those that increase it lead to excess.

Binder Syndrome

Perhaps one of the more subtle expressions of median facial deficiency is *Binder syndrome*, the syndrome of maxillonasal dysplasia detailed by von Binder in 1962⁴⁶ based on three cases. The anterior nasal spine is replaced by a pit in the maxilla known as the *fossa prenasalis*, which was first described by Zuckerkandl in 1882.⁴⁷ It is regarded as autosomal recessive with low penetrance.⁴⁸ Binder facies have been associated with chondrodysplasia punctata, and features such as brachydactyly should stimulate screening investigations.⁴⁹

Clinical Features

Patients with Binder syndrome have a typical facies with nasal flattening and malar hypoplasia. Nasal features are the consequence of a lack of foundation for the nose (absent anterior nasal spine and short septum) leading to flattened alae, an acute columellar-labial angle, and an abnormal nasofrontal angle (Fig. 18-7). A pathognomonic finding that differentiates this from other causes of midfacial hypoplasia is a lack of a palpable anterior nasal spine. Maxillary hypoplasia may be significant, leading to a class III skeletal relationship with anterior open bite. There may be a deficiency of nasal lining, the frontal sinuses may fail to pneumatize, and atlantoaxial defects



Fig. 18-7 Binder syndrome. A 1½-year-old girl with nasal hypoplasia. The lateral view shows a suggestion of relative prognathism.

may occur.⁵⁰ Radiographs are of little use in diagnosis, but a lateral cephalogram is important for documenting the occlusal relationship.

Principles of Correction

Given the psychosocial factors, nasal correction is often performed when the patient is approximately 10 years of age. The mildest forms can be addressed using standard rhinoplasty techniques; other methods include the use of a cantilevered bone or costochondral graft through a bicoronal approach, together with batten grafts to the alae. Medpor implants have been tried, but I avoid using alloplast in the nose because of the extrusion rate. If there is a lack of nasal tip support, a hinged septal flap to rotate the tip forward can be used; if there is a lack of bony dorsum, an L-shaped iliac bone graft can be used. Both may be performed through vestibular incisions and combined with onlay bone grafting around the piriform aperture to address the maxillary hypoplasia.⁵¹

Mild malocclusions can be treated with orthodontics alone; more significant abnormalities will require a combined approach. It is best to leave orthognathic correction until dentofacial growth is complete, and it should be noted that a standard LeFort I osteotomy will not improve nasal projection. However, if a significant skeletal class III relationship exists, a LeFort I osteotomy can be performed in the standard manner.

Median Facial Dysgenesis With Excess

The 0-14 cleft with excess is described as consisting of the cranium bifidum with duplication of the crista galli, orbital hypertelorism, nasal septal duplication, and a median cleft of the maxilla and lip.³⁴ It is very heterogeneous in its presentation and has many names, but is most commonly called *frontonasal dysplasia* (FND)⁵² (Fig. 18-8). It comprises some or all of the features above and a broad nasal root, with or without notching or clefting of the nasal alae. Soft tissue excess is manifest as a surplus of skin, hair (widow's peak), and fibrofatty tissue in the midline. Intracranially, median lipomas can also be observed, and in severe cases the median cranial base is disrupted by an encephalocele.



Fig. 18-8 Frontonasal dysplasia. Note the severe orbital hypertelorism and broad forehead with a widow's peak, together with widely spaced and notched nasal alae.

FNDs are a usually sporadic phenomenon of midline craniofacial excess, with few inherited cases. Other anomalies are common, but syndromic forms are rare and include acromelic frontonasal dysostosis, manifest as FND with tibial hypoplasia and preaxial polydactyly. It was hypothesized that mutations in the SHH pathway were responsible for this: *Gli3* is an important downstream target of SHH.^{53,54} Like FND, *orbital hypertelorism* is regarded as causally heterogeneous and is typically sporadic but a number of syndromes exhibit hypertelorism including Apert syndrome (*FGFR2*), Gorlin syndrome (*PTCH1*) and Greig syndrome (*GLI3*).⁵⁵

Genetic mutations have been identified within a family of homeobox genes resulting in a classification of some cases into types 1 through 3. Type 1 (frontorhiny) is a distinct phenotypic subset with autosomal recessive loss of function in the transcription factor *ALX3*.⁵ Type 2 FND has similar features, with abnormal skin and hair development, and is due to homozygous mutation in *ALX4*.^{56,57} In contrast to the *ALX3* form, it lacks the characteristic long philtrum with prominent ridges. Heterozygous mutations have been associated with parietal foramina.⁵⁸ Type 3 FND is caused by mutations in *ALX1* and exhibits a severe phenotype, with bilateral cleft lip and primary palate that are not seen in the *ALX3* and *ALX4* forms.⁵⁹ In contrast to *ALX3* and *ALX4*, *ALX1* is expressed in the early stages of neural crest development, and its dysfunction inhibits neural crest migration in the frontonasal stream.⁶⁰ Although *ALX4* may act downstream of the SHH pathway, how *ALX* genes interact with other signaling systems in craniofacial development remains to be determined.⁶¹ But knockdown of *alx1* leads to defective neural crest migration into the frontonasal and maxillary processes, indicating that this may underlie type 3 FND.⁶⁰

Craniofrontonasal Dysplasia

A distinct syndromic form of FND is *craniofrontonasal dysplasia* (CFND) in which orbital hypertelorism occurs in tandem with bicoronal craniosynostosis and a variety of extracranial abnormalities. It is inherited in a sex-linked fashion, but unusually, girls predominate and have a more severe phenotype. CFND is a consequence of a mutation of *ephrin-B1* (*EFNB1*) expressed in the frontonasal neural crest.^{62,63} Ephrin-B1 has a role in the proper migration of the cranial neural

crest, and disruptions lead to craniofacial malformations.⁶⁴ The developmental mechanisms of *EFNB1* remain to be elucidated, but there are indications that apart from signaling hierarchies, other factors that regulate cell proliferation, migration, and adhesion play a role in facial morphogenesis.

Clinical Features

All patients with CFND have unilateral or bilateral coronal synostosis, orbital hypertelorism, and other facial anomalies, such as CL/P, anterior open bite, hypoplastic ear deformities, and Stahl's ear. Eye abnormalities are related to oculomotor disorders and downslanting palpebral fissures. Dry, frizzy hair and longitudinal grooving of the nails are characteristics of patients with CFND. Limb anomalies such as clinodactyly, syndactyly, and brachydactyly are common, as is a characteristic shoulder girdle abnormality with neck webbing, prominent scapulas, and a hypoplastic clavicle. Management of these patients is the same as for others with syndromic craniosynostoses, with early frontoorbital remodeling to correct the calvarial morphology and orbital osteotomies to correct the hypertelorism after eruption of the permanent dentition.⁶⁵ Surgical correction is discussed in Chapter 25.

DEFECTS OF FACIAL FUSION: FACIAL CLEFTS

The facial processes consist of a core of neural crest–derived and somitomeric mesoderm (mesenchyme) enclosed in an ectodermal jacket. Each will have an associated nerve and vessel; the frontonasal process contains the ophthalmic nerves and vessels. The first arch vessel is the maxillary artery and the nerve is the trigeminal nerve with divisions to the respective maxillary and mandibular processes.

The argument of whether neural crest cells truly migrate was settled more than 20 years ago by techniques that can label individual cells and track their migration within live embryos in the laboratory.⁶⁶ Likewise, the mesodermal penetration versus fusion theory of how the facial processes coalesce points toward fusion as the mechanism. Proliferation of the mesenchyme leads to an increase in the size of the facial processes as they grow toward each other and fuse in a coordinated fashion. During the fifth gestational week the nasal placodes begin to sink into the frontonasal process, creating medial and lateral processes as distinct thickenings on either side of the developing nasal pit. At this stage the maxillary process fuses with the lateral nasal process; as they merge, the ectoderm in the floor of this groove forms a solid epithelial cord, which will eventually canalize to form the nasolacrimal duct, connecting the orbit to the nasal cavity. By the seventh week, the maxillary process almost reaches the midline, where it fuses with the paired medial nasal processes. The maxillary process will form the upper lip, upper jaw, and secondary palate, whereas the philtrum and primary palate are derived from the medial nasal process; the lateral nasal process forms the ala of the nose. Meanwhile, the mandibular process grows medially to join the fellow of the opposite side to complete the formation of the lower jaw. The maxillary process also fuses longitudinally to the mandibular process; the degree of this fusion determines the width of the mouth.

Signaling Networks Control Outgrowth and Patterning of the Facial Processes

The growth and patterning of the facial processes are tightly regulated by numerous signaling pathways and regions that control epithelial-mesenchymal interactions, cell proliferation, and cell differentiation. Members of the TGF β subgroup called the *bone morphogenetic proteins* (BMPs), *Bmp2* and *Bmp4*, are important for outgrowth of the facial processes. Ectopic application can lead to duplicated structures, increased cell proliferation, and activation of the downstream genes

Msx-1 and *Msx-2*.⁶⁷ By contrast, inhibition leads to decreased proliferation and apoptosis.⁶⁸ Work has shown that the BMPs can alter the shape of the frontonasal process and create species-specific differences in morphology. Similarly, FGF signaling around the nasal pits is important, with decreased FGF signaling leading to a failure of contact between the processes, resulting in failure of fusion.³³ The BMPs act upstream of the FGFs, and the *Msx* genes are the downstream effector molecules for both pathways.³³ *MSX-1* mutations have been associated with nonsyndromic CL/P and syndromic CL/P with oligodontia.^{69,70} Nonsyndromic CL/P associations have also been reported with multiple FGFs and supported by mouse models and may constitute up to 3% to 5% of nonsyndromic CL/P cases.^{71,72}

The *Wnt signaling molecules* (the name comes from *wingless* in *Drosophila* and *integrated-1* [*int1*] in mice when they were found to be the same gene) are secreted signaling molecules, and a number of Wnts are known to be expressed in the developing facial processes. Particularly, *Wnt3* is expressed in the facial ectoderm of the tip of the maxillary and medial nasal (but not lateral nasal) process.⁷³ *WNT3* homozygous loss results in cleft lip only and amelia.⁷⁴ *WNT9a* is close to *WNT3* on the chromosome and is expressed in the tips of all the facial processes but not the secondary palate. Although further investigation is necessary to dissect how the *Wnt* genes effect facial morphogenesis, this pathway appears important in mediating FGF signaling loops and interacts with the BMP pathway.

Fusion of the facial processes involves a series of morphogenetic mechanisms:

- As the processes draw near, the periderm (outer layer of the bilaminar fetal epidermis) is shed, mediated by apoptosis; it is suggested that this promotes cell junction adhesion between the two epithelia.⁷⁵
- Thereafter, epithelial filopodia at the primary site of fusion begin to establish bridges between the processes resulting in an epithelial seam. This breaks down to allow confluence of the underlying mesoderm. This mechanism has been primarily studied in the secondary palate, the key debate being whether these cells undergo programmed cell death or transition into mesenchymal cells.⁷⁶

Evidence so far suggests that the cells undergo apoptosis, whereas perhaps a small population undergoes epithelial-mesenchymal transition.⁷⁷ Failure of fusion leads to facial clefts, and this can be caused by failure of one of the following:

- Neural crest migration
- Cell proliferation
- Adherence between processes
- Programmed cell death during fusion
- The normal morphogenetic processes after initial stages of fusion

All these processes are under genetic and epigenetic control.

Clefts of the upper lip (including Tessier 1 and 2 clefts) occur as a consequence of a failure of normal fusion of the maxillary and medial nasal processes. Cleft lip may be associated with cleft palate (discussed later) and may be syndromic and nonsyndromic. By contrast, atypical facial clefts are considered sporadic phenomena that are etiologically heterogeneous. Defects in the fusion of the lateral nasal and maxillary processes lead to the oroocular cleft (Tessier 3). Insufficient fusion between the maxillary and mandibular processes causes macrostomia (Tessier 7 cleft). Clefts 6 and 8 are actually hypoplastic disorders discussed later.

The Problem of the Tessier 4 and 5 Clefts

Tessier clefts 4 and 5 of the face do not map to known fusion sites. This poses a problem to explain in simple embryologic terms. It may be that these are caused by amniotic bands, but be-

cause they occur in a relatively predictable fashion, this seems unlikely. They may be caused by external pressure or internal hemorrhage, leading to tissue necrosis. Another theory may be that there is a failure of fusion of the maxillary and lateral nasal processes with a degree of embryonic regulation occurring. Embryonic regulation is the initiation of reparative mechanisms when part of the embryo is lost by trauma or experimental manipulation. Given the discrete signaling centers around the facial processes, it is possible that a degree of incomplete regulation occurs leading to these clefts.

Another possibility is that the problem of the Tessier 4 and 5 clefts may be manmade, in that the classification system attempts to classify a diverse range of craniofacial morphologies into a limited number of groups. There is a tendency in the literature to make the embryology fit the classification rather than the reverse. Each cleft is unique and a consequence of numerous mechanisms, such as failure of proliferation, failure of fusion, mechanical effects, and regulation.

DEVELOPMENT OF THE PALATE

The oronasal cavity begins as a single large cavity, but between the six and tenth weeks of gestation, the formation of the palate partitions it into separate spaces. The palate is derived from the frontonasal process that forms the primary palate, which constitutes the anterior bony part of the upper jaw (the premaxilla), which bears the incisor teeth. Paired lateral palatine processes positioned on the oral surface of the maxillary process appear at the sixth week of development and form the secondary palate. Cell proliferation causes them to grow medially and inferiorly down either side of the tongue. During the seventh week, over a period of hours, these palatine shelves elevate to achieve their anatomic position above the tongue.

Palatal Elevation

A key event in palatogenesis is the elevation of the palatal shelves. Most theories invoke an intrinsic force within the shelves that overcomes extrinsic forces and culminates in palatal shelf elevation. Studies have implicated hyaluronic acid as a likely mediator; this glycosaminoglycan carries a high negative charge that attracts water, creating an amorphous gel.⁷⁸ The increase in hyaluronic acid causes the palatal shelves to swell, and this swelling force is directed by longitudinal bundles of type I collagen oriented from the base to the shelf edge. Palatal mesenchyme cells are contractile and secrete neurotransmitters, which may control contractility and glycosaminoglycan degradation, thereby playing a modulatory role in palatal morphogenesis.⁷⁹

During the period of shelf elevation, there is an increase in head height but not head width, creating space above the tongue into which the shelves move. The tongue muscles become functional about the time of shelf elevation, and conversely, inhibition of fetal movement has been suggested to cause palatal clefting.⁸⁰ Descent of the tongue is a key event, which if restricted by mandibular hypoplasia such as in Pierre Robin sequence, may result in cleft palate. Although the exact combination of mechanisms remains to be fully characterized, it appears that an intrinsic elevation mechanism of the shelves in concert with extrinsic environmental factors and mandibular morphogenesis allows the appropriate temporospatial sequence of events.

Palatal Fusion

The *medial edge epithelia* (MEE) of the fusing shelves forms a midline seam, and then rapidly disappears to allow mesenchymal continuity. During this period, the ectoderm on the nasal surface differentiates into epithelium characteristic of the respiratory tract, while the oral surface forms

the nonkeratinizing oral mucosa. Within the mesenchyme, the palatal shelves will ossify to form the hard palate, while myoblasts migrate into the posterior palate to contribute to the soft palate. Palatal fusion is complete by 12 weeks.

Molecular Signaling and Palate Development

Much of our knowledge of molecular signaling and palate development is based on the mouse model using targeted mutagenesis or manipulation of the palatal shelves in explant culture. SHH is present in much of the early oral epithelium and is a key signal in promoting palatal shelf outgrowth.⁸¹ SHH in palatal explant cultures stimulated cell proliferation. *Fgf10*, secreted by the palatal mesenchyme together with its receptor *Fgfr2*, in the overlying epithelium are also critical for palatal shelf outgrowth. *Fgf10* knockout mice have cleft palate with hypoplasia as do those with targeted mutation of *Fgfr2* in epithelium.⁸² In embryos with loss of *Fgf10* or *Fgfr2*, there was a drop in SHH expression indicating that *Fgfs* may act through SHH to effect cell proliferation and palatal shelf growth. SHH may act through the BMP pathway: SHH protein beads activate *Bmp2* expression in culture, and *Bmp2* can increase cell proliferation in the palatal mesenchyme.⁸³ The *Bmp* signaling regulates expression of the transcription factor *Msx1* and is also responsible for maintaining SHH expression in the palatal epithelium.

In the mouse model, there is a degree of regionalization along the rostrocaudal axis of the morphologically homogeneous palatal shelves. For example, *Msx1* is expressed anteriorly whereas *Tbx22* is expressed posteriorly in the presumptive soft palate mesenchyme. *Tbx22* appears to be required for palatal bone formation with *Tbx22* null mutants exhibiting cleft palate or submucous cleft palate (SMCP).⁸⁴ Mutations in *TBX22* have been identified as a cause of X-linked syndromic cleft palate with ankyloglossia termed CPX.⁸⁵

In addition to the rostrocaudal patterning, there are signals that are involved in mediolateral patterning of the shelves (note that the medial surface becomes the nasal surface of the shelf after elevation). *Dlx5* is coexpressed with *Fgf7* in the medial mesenchyme of the palatal shelf. Loss of *Dlx5* leads to a loss of *Fgf7* and an expansion of the SHH expression onto the medial epithelium (Fig. 18-9). Such embryos had an expanded oral aspect to the palate and a malformed soft palate.⁸⁶

p63 is an ectoderm-specific p53-related transcription factor that acts upstream of *Dlx5* and *Dlx6*, regulating their expression. p63 regulates morphogenesis of surface ectoderm through multiple signaling pathways.⁸⁷ Mutations in p63 are responsible for *ectrodactyly ectodermal dysplasia clefting syndrome* (EEC), which is an autosomal dominant syndrome comprising a cleft hand and abnormal skin, teeth, and hair, together with CL/P. p63 binds to an enhancer upstream of *IRF6* (interferon regulatory factor 6), which is a direct target of p63.⁸⁸ *Irf6* is expressed at the edges of the fusing palatal shelves and facial processes. Loss of *Irf6* leads to a hyperproliferative epidermis that fails to differentiate, causing cleft palate and mirroring a number of ectodermal dysplasia syndromes.⁸⁹ Compound mutants of p63 and *Irf6* exhibited cleft palate with retention of the periderm cells obstructing the fusion of the palate. *IRF6* mutations have been identified in Van der Woude syndrome⁹⁰ and may have a role in nonsyndromic clefts.⁹¹

TGFβ3 has a prominent role in fusion of the palatal shelves. Palatal shelves in *TGFβ3*^{-/-} mutant mice elevate and make contact but fail to fuse with persistence of the MEE. Both *TGFβ1* and *TGFβ3* are expressed in the MEE, and there is a degree of redundancy, because a knockin of *TGFβ1* into the *TGFβ3* locus partially rescues the cleft phenotype. In addition, a significant reduction in programmed cell death was detected, indicating that *TGFβ3* regulates apoptosis in the MEE.⁹²

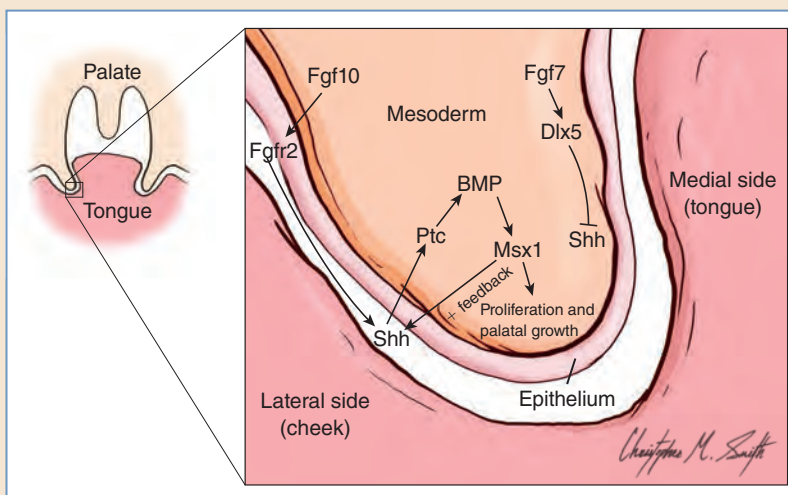


Fig. 18-9 Signaling in the palate. Palatal development involves cellular proliferation with growth of the palatal shelves down the side of the tongue, and elevation and fusion in the midline. *FGFs*, *SHH*, *BMPs*, and *Msx* genes are part of a signaling network that control growth of the palatal shelves. Additionally, *FGFs* through the action of the transcription factor *Dlx5* act to suppress *SHH* and specify a medial-lateral identity to the palate. Note that the medial surface of the palate becomes the nasal surface after elevation of the palatal shelves. (*BMP*, Bone morphogenetic protein; *Fgf*, fibroblast growth factor; *Ptc*, patched; *Shh*, Sonic hedgehog.)

Genetic and Environmental Factors in Palatal Clefts

Any failure in the timing or mechanism of these many processes may result in cleft palate, so it is not surprising that CL/P is one of the most common craniofacial malformations. The basis of nonsyndromic clefting is not easily explained by a single gene model, and it was thought that a multifactorial threshold model was likely; for example, after a certain number of mutations occur in the genetic background, a phenotype is seen. In this situation, a greater number of mutations would result in a more severe phenotype and a higher risk of recurrence. However, a large Norwegian population study did not find this association, and new models need to be found.⁹³ Environmental factors include the mother's use of anticonvulsant medications, steroidal agents, alcohol, smoking, and an excess of retinoids (vitamin A); folate deficiency has been implicated as well, whereas the genes associated with sporadic CL/P include *TGFβ3*, *IRF6*, and *MSX1*. Increasingly, the importance of gene-gene and gene-environment interactions is being realized; for example, it has been demonstrated that smoking and alcohol use in the presence of *MSX1* mutations have a synergistic effect in causing isolated cleft palate (iCP).^{94,95}

A number of genes have been identified for syndromic CL/P and iCP, such as Apert syndrome, for which cleft palate is more commonly related to the S252W mutation in *FGFR2*, possibly altering the balance of *FGFR2* expression and events at the MEE. A targeted point mutation in *Fgfr2*^{C342Y/-/-} that leads to ligand-independent activation of the receptor results in increased lateral palatal mesenchyme proliferation, reduced GAG accumulation, and a delay in elevation.⁹⁶

SYNDROMIC CLEFT PALATE

CL/P and iCP are discussed in Chapter 22; however, the more common syndromes are reviewed next.

Pierre Robin Sequence

Defined as a triad of micrognathia, glossoptosis, and airway obstruction (Fig. 18-10), *Pierre Robin sequence* is a well-recognized clinical entity, but variable in terms of presentation and diagnostic criteria.⁹⁷ It may affect up to 1:8500 births.⁹⁸ More than 90% of cases exhibit cleft palate that is characteristically wide and U-shaped. The sequence is hypothesized to be that reduced mandibular growth leads to a tongue that fills the oronasopharynx and physically obstructs the elevation of the palatal shelves. Affected babies have significantly smaller mandibles and tongues than age-matched controls at birth, but significant increases in size dramatically enlarge the airway, and by 2 years of age this is manifested as an improvement in airway control and feeding.⁹⁹ More than 40 syndromes with Pierre Robin sequence have been described; the most common include Stickler syndrome and velocardiofacial syndrome (22q). Although some genetic mutations have been reported, no specific gene causes Pierre Robin sequence.¹⁰⁰

Management

Airway Airway obstruction is a defining feature of Pierre Robin sequence, and although there is posterior displacement of the tongue into the hypopharynx, the airway may be compromised at many levels, including central abnormalities, and pharyngeal and laryngeal anomalies. Airway compromise leads to growth retardation, decreased feeding, and increased energy expenditure from the work of breathing; management of airway obstruction is the same as for any craniofacial patient. Bedside endoscopic assessment to evaluate the degree and level of obstruction is recommended, but this may require examination under anesthesia and direct laryngoscopy and bronchoscopy to rule out multilevel obstruction and guide intervention. The airway improves in the majority of cases in the first year, which is attributed to mandibular catch-up growth.



Fig. 18-10 Pierre Robin sequence. Mandibular hypoplasia is observed, with a wide complete cleft of the secondary palate. This infant demonstrates respiratory compromise, as evidenced by sternal retraction. Note the feeding tube in place to ensure that adequate nutrition is provided despite airway issues.

Surgical management of the Pierre Robin sequence airway is usually not required, but, when performed, centers on the use of tongue-lip adhesion, and more recently, mandibular distraction. Tongue-lip adhesion involves the anterior anchoring of the posterior tongue with a nonabsorbable circummandibular suture and the anterior tongue to the lower lip mucosa. There has been debate about the benefit of this procedure with regard to improved feeding, because it does disrupt the normal swallow mechanism. A GILLS scoring system to guide patient selection has been proposed that is based on the following¹⁰¹:

- Gastroesophageal reflux
- Intubation before surgery
- Late presentation
- Low birth weight
- Syndromic diagnosis

Mandibular distraction is specifically targeted to the obstruction caused by the tongue base and aims to increase pharyngeal airway diameter. The principles of distraction are discussed in Chapter 30. Internal or external distractors can be used; the procedure involves bilateral mandibular osteotomies to lengthen the body of the mandible. Potential complications include damage to tooth germs, nerve injury, wound complications, and malunion. Large-scale studies have yet to report their outcomes, but objective radiographic studies have demonstrated increases in airway size; overcorrection is recommended.⁹⁷

Feeding Maintaining adequate respiratory stability during feeding is essential. This is mainly accomplished by using modified bottles or nasogastric tube feeding. Nasogastric tube feeding is rarely required after 1 year of age in isolated Pierre Robin sequence cases; children with prolonged issues or requiring gastrostomy tube feeding are usually those with associated syndromes.

It is critical to monitor the infant's weight gain. Failure to gain weight should prompt investigation and intervention. Reflux can unbalance feeding and respiration by causing airway inflammation and edema, increasing secretion production and compromising the swallow mechanism. It is important that reflux be considered, identified, and treated early on to optimize feeding.

Further Assessments All Pierre Robin sequence babies should have an ophthalmologic assessment between 6 and 12 months or at the time of a molecular genetic diagnosis. Newborns are hyperopic, so any myopia at 6 to 12 months should raise suspicion of Stickler syndrome; although not quantified, attention tests are used to pick up early visual problems. Other investigations are geared toward searching for features of other known syndromes, assessment of hearing, and monitoring speech development.

Stickler Syndrome

Stickler syndrome is an autosomal dominant mutation of *Col2a1*, *Col11a1*, or *Col11a2*, with incomplete penetrance called *types 1, 2, and 3*, respectively. They are procollagen genes that are expressed in the cartilage and vitreous, thereby affecting the development of the mandible and eyes. The syndrome comprises Pierre Robin sequence features, eye defects, progressive sensorineural hearing loss, joint hypermobility or hypomobility, and variable epiphyseal dysplasia (manifested as joint pain, degeneration, or dislocation).

Eye Defects

Eye defects comprise a progressive severe myopia (80%), vitreous degeneration, and the most emergent problem, retinal detachment. There should be a high index of suspicion for retinal detachment. Babies and young children will not declare visual problems until the second eye becomes involved; thus ophthalmologic screening should be performed in confirmed cases. Retinal tears and detachment occur peripherally at first, then intrude on central vision. A careful family



Fig. 18-11 This 13-year-old boy with Stickler syndrome was born with micrognathia and a wide complete cleft of the secondary palate. He is myopic and has additional ophthalmologic findings consistent with Stickler syndrome. He had a tracheostomy for severe airway compromise and associated sleep apnea, which followed a pharyngeal flap. After takedown of the flap and additional mandibular growth, he was able to be decannulated. He wears bilateral hearing aids.

history from the parents regarding eye problems may suggest Sticker syndrome in a child with cleft palate. It has been suggested that prophylactic 360-degree cryoretinopexy will reduce the risk of detachment later in life.¹⁰² Many patients develop visual loss from progressive glaucoma.

Clinical Features

Facies include maxillary hypoplasia and dental issues such as malocclusion, natal teeth, and enamel hypoplasia. Progressive high-frequency sensorineural hearing loss is present in 80% of children with Stickler syndrome, but symptoms are not seen until the third decade (Fig. 18-11). All affected individuals have musculoskeletal anomalies, including mild to moderate epiphyseal dysplasia. Many develop premature joint disease, especially of the hip, and spinal deformities also occur.

Management

Patients with Stickler syndrome are managed in the same way that Pierre Robin sequence babies are managed, with ophthalmic screening. Abnormalities should prompt referral to a genetics specialist, and referral to an orthopedic surgeon may be considered.

Van der Woude Syndrome

Van der Woude syndrome is an autosomal dominant disorder of orofacial anomalies with variable penetrance that commonly manifests as CL/P and distinctive lower lip pits. It is the most common form of syndromic CL/P, affecting 1 to 2:100,000 live births, or 1% to 2% of CL/P patients. There is no racial or sex predilection, and 25% have minimal findings, such as absent teeth or lower lip pits. An accurate family history is essential to diagnosis, but 30% to 50% have de novo mutations; the examiner should look for an autosomal dominant inheritance, bearing in mind the variable penetrance. The fact that individuals may have CL/P or iCP indicates that these conditions are not entirely independent.



Fig. 18-12 Van der Woude syndrome. This is the classic position of the distinctive lower lip pits associated with this syndrome. This child was not troubled with serous discharge from these well-developed sinuses.

Van der Woude syndrome has been identified as a 1q32 deletion, and a number of mutations in *IRF6* have mapped to this locus.¹⁰³ It is part of a family of helix-turn-helix transcription factors that control interferon; it is expressed along the edges of the fusing palate and facial processes, but the function of *IRF6* remains unknown.

Clinical Features

The hallmark features of Van der Woude syndrome are the CL/P and lower lip pits (in 70% of patients) (Fig. 18-12). The lower lip pits typically sit on either side of the midline, but aberrant positions have been described. They may communicate with minor salivary glands and hence secrete saliva. Excision of the pits must therefore include the surrounding glandular tissue to prevent mucocoele formation. Lower lip pits exist in 2% of the normal population and may be associated with preauricular sinuses. SMCP is common and should be excluded, and it is important to search specifically for the triad of features and speech assessment for hypernasality. Extraoral anomalies include limb anomalies and popliteal webs; in this instance, it is called *popliteal pterygium syndrome* and is also caused by mutations in *IRF6* in 97% of cases (rather than the 76% of Van der Woude cases).

DEVELOPMENT OF THE PHARYNGEAL ARCHES AND BRANCHIAL ARCH SYNDROMES

The *pharyngeal arches* are a segmental series of structures in the cephalic region from which the nerves, muscles, and skeletal components of the pharyngeal apparatus are derived. Externally, between each arch lies a pharyngeal cleft; internally, there is the equivalent pharyngeal pouch. Each arch consists of mesenchyme that is mainly derived from the neural crest and some pre-muscle mesoderm that has migrated from the somitomeres. The generic constituents of each arch are a cartilage rod, an aortic arch, a nerve, muscle, and derivatives of the arch endoderm.

The first pharyngeal arch is called the *mandibular arch*, yet it also gives rise to the maxillary processes. Its skeletal component is Meckel's cartilage, which gives rise to the sphenomandibular ligament, malleus, and incus. The aortic artery of this arch will become the maxillary artery, and the muscles of this arch are all innervated by the trigeminal nerve. The second arch is called the *hyoid arch*, and its skeletal component forms the lesser horns and body of the hyoid, stylohyoid ligament, styloid process, and stapes. The muscles of facial expression are derived from this arch mesoderm and are innervated by the facial nerve. The stapedial artery supplies this arch; its course passes through the stapes, and regression during the third fetal month creates the unique morphology of this bone. This artery may persist into adult life, where it is at risk during middle

ear surgery. Gestational hemorrhage from this artery has been cited as a cause for some branchial arch anomalies, such as hemifacial microsomia.¹⁰⁴

The first pharyngeal cleft gives rise to the external auditory meatus, whereas the remaining clefts are covered by a downward growth of the second arch called the *operculum*, and the space underneath is called the *cervical sinus*. This eventually expands into the smooth contours of the neck as development proceeds. The third arch gives rise to the internal carotid artery and the greater horn of the hyoid, with its only muscular derivative being the stylopharyngeus, innervated by the glossopharyngeal nerve. The fourth and sixth arches are less well studied, but the fourth arch gives rise to the muscles and cartilages of the larynx and lower pharynx; these are innervated by the vagus nerve. The arteries of the fourth arch form the right subclavian artery and aorta. The sixth arch may contribute to the laryngeal and tracheal cartilages; the fifth arch does not form in higher mammals.

The endoderm lines the internal aspect of the pharyngeal arches. Early in development, the pharyngeal endoderm begins to evaginate toward the ectoderm and assumes a slitlike morphology under the constraining influence of a web of actin cables. The endoderm of these pouches gives rise to the thyroid, thymus, and tonsil. At the dorsalmost part of the pouch, endodermal *Bmp7* signals to the ectoderm of the pharyngeal cleft to induce the epibranchial placodes.¹⁰⁵ These placodes are thickenings of the ectoderm at the dorsal-caudal rim of the arch that will produce neuroblasts, which migrate and condense to form the cranial nerve sensory ganglia. The first pharyngeal pouch deepens and expands to form the middle ear cavity, while its connection to the pharynx persists as the Eustachian tube. Commensurate with this, the first pharyngeal cleft elaborates a cord of epithelium that canalizes, creating the external auditory meatus. Between the cleft and the pouch persists a membrane, the *membrana tympani*.

Signaling Systems Pattern the Arches to Establish Normal Facial Morphology

Each pharyngeal arch gives rise to a range of structures, each of which is different from its neighbor. Therefore every arch requires a positional identity to 'know' where it is and what to form. Initially, it was thought that all this information was carried into the arch by the migrating neural crest, but a role for the pharyngeal endoderm has become apparent.^{106,107} The pharyngeal endoderm appears to be the source of signals that establish pattern in the pharyngeal arches; for example, specific combinations of homeobox genes in the first arch are responsible for the different tooth types.^{108,109}

In the first arch, SHH from the endoderm appears to be important in establishing the localized expression of *Fgf8*. Experimental manipulation of SHH expression can reduce or increase the *Fgf8* expression, resulting in abnormal patterning of the lower jaw.¹¹⁰ *Fgf8* plays a role in restricting *Bapx1* expression to a specific area of the first arch. Excess *Fgf8* inhibits *Bapx1* and leads to loss of the jaw joint. Manipulating the expression pattern of *Bapx1* leads to changes in development and position of the jaw joint.¹¹¹ *BAPX1* abnormalities have been identified in some patients with oculo-auriculo-vertebral syndrome (hemifacial microsomia).¹¹²

In the search for animal models with abnormal pharyngeal arches, a zebrafish mutant called *van gogh* fails to form pharyngeal pouches. Accordingly, when the otherwise-normal neural crest streams reach the arches, they become disorganized, and abnormal skeletal structures form.¹¹³ *van gogh* was found to be a loss of function of *tbx1*, a T-box transcription factor strongly implicated in DiGeorge syndrome.¹¹⁴ In mouse, knockout of *Tbx1* results in a 22q deletion-like phenotype, whereas mutations in *TBX1* have been identified in individuals with 22q DS.¹¹⁵⁻¹¹⁸ Little is known about the mechanism of *Tbx1* or its gene targets, although the *Fgfs* are candidates. *Fgf8* knockouts have features similar to *Tbx^{-/-}* mice, with normal *Tbx1* expression indicating that *Fgf8* lies downstream in the signaling pathway. By contrast, Wnt signaling through β -catenin appears to lie upstream of *Tbx1*.¹¹⁹⁻¹²⁰

22q Deletion Syndrome (Velocardiofacial Syndrome/DiGeorge Syndrome)

22q deletion syndrome (22q DS) was described by Shprintzen et al¹²¹ based on the assessment of 12 children with similar anomalies, all of which referred for velopharyngeal insufficiency. It comprises a “CATCH-22” of anomalies (Fig. 18-13):

Cardiac anomalies

Abnormal facies

Thymic aplasia

Cleft palate

Hypocalcemia

22q DS shares many features with DiGeorge syndrome, and both were noted to possess a chromosome deletion at 22q; this (heterozygous) deletion encompasses at least 32 genes, including *TBX1*.¹¹⁷⁻¹²² Other genes in the 22q DS region may also cause the characteristic phenotype by acting as genetic enhancers or modifiers, but *TBX1* mutations in patients that are 22q positive also

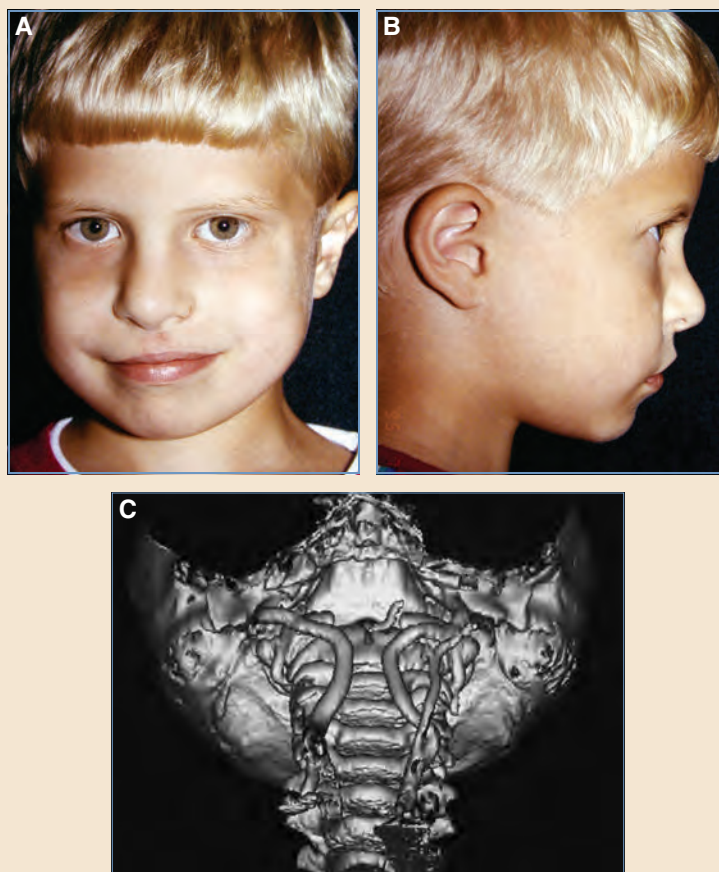


Fig. 18-13 22q deletion syndrome. **A** and **B**, This 7-year-old boy had been receiving speech therapy since he was 3 years of age. He has a short, immobile soft palate with a submucous cleft. He exhibits a broad nasal dorsum with narrow alar bases, flattened zygomatic arches, platybasia, and vertical maxillary excess. At the time of his planned pharyngoplasty, it was noted that he had medially displaced carotid arteries. **C**, CT angiography of the carotids of another patient with the same syndrome.

replicate the characteristics of the syndrome and make *TBX1* a key suspect in the pathogenesis of 22q DS. DiGeorge syndrome affects 10% of 22q DS patients: this is a developmental defect that additionally involves the third and fourth pharyngeal pouches with thymic aplasia and hypoparathyroidism manifested as hypercalcemia.

Clinical Features

Velopharyngeal Insufficiency Twenty-four percent of these patients present with iCP or SMCP, which is characterized by the triad of notched posterior hard palate (absence of posterior nasal spine), a bifid uvula, and zona pellucida because the muscle fails to meet at the midline of the palate. These individuals have a limited ability to open their mouths, adenoidal hypoplasia, and platybasia. Platybasia creates a deep pharynx with poor lateral wall movement that contributes to the velopharyngeal insufficiency, and it also makes access difficult for pharyngeal flap surgery. Speech outcomes are typically poorer as a result of anatomic and neurocognitive learning deficits. The cleft, often compounded by patent Eustachian tubes, predisposes the patient to middle ear disease and consequent hearing impairment, but sensorineural hearing loss may also occur.

Cardiac Anomalies Cardiac anomalies are present in 85% of patients with craniofacial anomalies, including ventriculoseptal defects (most common), aortic coarctation, pulmonary stenosis, and tetralogy of Fallot. Such conotruncal cardiac defects may present with recurrent chest infections, compounded by immunodeficiency in the DiGeorge group. Craniofacial surgeons must be aware that up to 25% of patients with velopharyngeal insufficiency have medially deviated or midline carotid arteries.¹²³ Before starting surgery, one should palpate the pharyngeal walls for pulsations to avoid raising pharyngeal flaps immediately adjacent to the vessels. The pharynx may be too deep to palpate the carotids reliably, and some surgeons advocate a CT or MR angiogram of the carotids to determine their proximity to the surgical site.

Facial Features Facial features characteristic of these patients include a long facial shape with vertical maxillary excess, flat malar eminences, and a broad nasal bridge. There may be mild epicanthal folds or pseudohypertelorbitism. The temporomandibular joint is relatively displaced posteriorly, leading to a class II occlusion. Such features are difficult to discern at birth.

Associated Factors Associated factors include mild hypotonia in early childhood, small stature, learning difficulties, psychosis in later life, immune issues from thymic aplasia, and abnormal dermatoglyphics.

Management

The palate is repaired in the standard fashion, but outcomes are poorer and the need for revision surgery is more common than in nonsyndromic cases. Careful assessment of the palate to inform selection of the primary procedure can produce results close to a nonsyndromic group, but these children require more intense speech therapy and take much longer to achieve such results.¹²⁴ Children with 22q DS should be seen by an immunologist to be assessed for immune compromise; until this occurs, they should not receive live vaccines. If they are not immune competent, the blood products they receive should be irradiated. Individuals with DiGeorge syndrome should have annual thyroid and calcium assessments and a CBC.

Oculo-auriculo-vertebral Spectrum (Hemifacial Microsomia and Goldenhar Syndrome)

Commonly termed *hemifacial* (craniofacial) *microsomia* and known by many other names, *oculo-auriculo-vertebral spectrum* (OAVS) is a syndrome that is characterized by a variable constellation of features related to the maldevelopment of the branchial arches.^{125,126} It is the most common craniofacial syndrome after CL/P, with an incidence of approximately 1:5600.¹²⁷ OAVS is bilateral in 10% to 20% of cases and has a male-to-female ratio of 3:2 and a right-to-left ratio

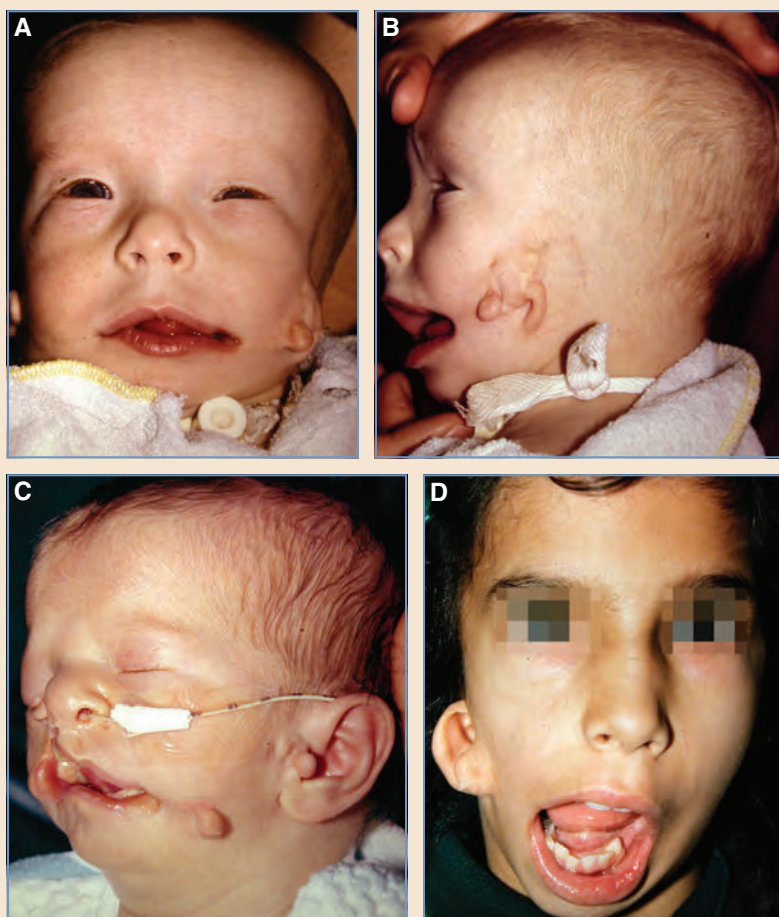


Fig. 18-14 Goldenhar syndrome presents in a spectrum ranging from mild to severe. **A** and **B**, This patient has hypoplasia of the soft tissue, mandibular hypoplasia, macrostomia, microtia, and epibulbar dermoid. **C**, A patient with Goldenhar syndrome with bilateral deformities, including asymmetrical mandibular hypoplasia, bilateral macrostomia, incomplete bilateral cleft lip (occult on the left side), nasolabial clefting, and preauricular remnants. **D**, A 9-year-old child with mandibular hypoplasia, auricular remnant, soft tissue deficiency, right macrostomia, and epibulbar dermoid.

of 3:2. The phenotypic subset known as *Goldenhar syndrome* (Fig. 18-14) constitutes approximately 4% of this group.¹²⁸ No minimum diagnostic criteria have been agreed on, and it is widely accepted that preauricular malformations and microtia represent microforms of the condition. Up to one third of cases may be bilateral. Microtia may be the only sign of OAVS in the early years, until mandibular hypoplasia becomes more apparent.

OAVS is a sporadic disorder, but there is some evidence for some inheritance with autosomal dominant inheritance in 2% to 10% of cases.¹²⁹ The inherited form is more likely to have bilateral involvement, but the patient is less likely to have hearing loss, epibulbar dermoids, and abnormalities of the external auditory meatus. The etiologic mechanism of OAVS has yet to be determined; hemorrhage of the stapedia artery has been suggested, based on animal models and case reports.¹³⁰ Numerous associations with maternal conditions and medications including thalidomide and retinoids have also been noted, suggesting that epigenetic mechanisms are

important. It should be noted that dysregulation of *BAPXI* has been reported, probably as a result of epigenetic factors.¹¹² This explains the high proportion of discordant monozygotic twins, why other genes are implicated, and the high correlation with maternal IVF treatment.¹³¹ It is also interesting to note that a gene that is critical for the position and development of the jaw joint is implicated in OAVS.¹¹¹

The underlying issue is a tissue deficiency. It remains to be seen whether the tissue deficiency is caused by a failure of neural crest specification, migration, proliferation, or abnormal signaling in the first arch or whether it is caused by cell death (either programmed or secondary to vascular trauma).

Clinical Features

The clinical features of oculoauriculovertebral spectrum syndromes are variable and wide ranging and are best summarized by the OMENS and OMENS+ classifications.^{132,133} The acronym stands for **o**rbit, **m**andible, **e**ar, **n**erve, and **s**oft tissue. The + denotes extracranial anomalies, and a phenotypic assessment tool exists to aid standardization of assessment.¹³⁴

Orbital Dystopia The orbit may be of abnormal size, abnormal position, or both; this is a result of hypoplasia of the maxilla and zygoma. In the Goldenhar subset, epibulbar dermoids are situated on the sclera, typically in the inferolateral part of the eye; these are masses of gray-white to yellowish tissue and vary in size. Lipodermoids, which are mobile yellow masses, are less common. The presence of epibulbar dermoids may indicate other eye anomalies such as ptosis, lacrimal drainage issues, and motility disorders.¹³⁵

Mandibular Hypoplasia Mandibular hypoplasia is regarded as the cornerstone of the disorder, because it is directly correlated with the severity of the other features.¹³² It leads to malocclusion and deviation of the chin to the affected side with mouth opening (Fig. 18-15). The hypoplasia was originally classified by Pruzansky,¹³⁶ was modified by Vento et al,¹³² and was assimilated into the OMENS classification guiding surgical management:

M1 is characterized by a small mandible with a short ramus and an intact glenoid fossa.

M2 indicates a small and abnormally shaped ramus and is subdivided:

Type A: Symmetrical glenoid fossae

Type B: A severely hypoplastic condyle that sits in an abnormal position

M3 has an absence of the temporomandibular joint.

M1 and M2A may be addressed with distraction techniques, whereas M2B and M3 require reconstruction, typically with costochondral rib grafts.

Ear Anomalies Preauricular tags, pits, and sinuses may occur, and the external auditory meatus may be malpositioned and hypoplastic. The relationship of the facial nerve may be abnormal, and this is important in surgical planning. Conductive hearing loss is common as a consequence of middle ear anomalies. By contrast, the inner ear is rarely affected. The auricular morphology was described by Meurmann and incorporated into the OMENS classification:

E1: There is hypoplasia, but all structures are present.

E2: There are remnants of cartilage and skin, with external auditory meatus atresia.

E3: The pinna is almost completely absent; the lobule usually remains in an anterior and inferiorly displaced position.

Nerve Defects The facial nerve is most commonly affected. It may have an unusual course or exhibit paresis, and the parotid nerve may be absent. Facial palsy is estimated to occur in 22% to 45% of patients with 22q deletion syndrome and may be caused by neural or muscular hypoplasia. Most facial nerve grading systems cannot be applied accurately, because the asymmetry at rest is partly the result of dentoskeletal factors. Other nerves may be smaller, such as the motor nerve to the masseter muscle; this should be taken into consideration when it is used to drive a free functioning muscle transfer.



Fig. 18-15 Hemifacial microsomia. This child has type III mandibular deformity, soft tissue hypoplasia, and lobule-type microtia.

Soft Tissue Defects Macrostomia (Tessier 7 cleft) is common, often minor, and is easily missed. Cleft palate is also common, especially in individuals with Goldenhar syndrome; the palate may be hypoplastic and deviate on examination; about one third of patients may have velopharyngeal insufficiency.¹³⁷

Other Anomalies Spinal anomalies, especially those affecting the cervical vertebrae, are common, such as C2-3 fusion, hemivertebrae, spina bifida, and craniocervical base anomalies. Such intracranial anomalies as hydrocephalus and intracranial lipomas as well as developmental delay may also occur.¹³⁸ Other defects include congenital heart disease, pulmonary anomalies, genitourinary malformations, and musculoskeletal defects.¹³³

Management

As with all complex craniofacial conditions, assessment and treatment should occur within a craniofacial team. In a neonate, multidisciplinary management with optimization of the airway and feeding is vital to define the extent of the disorder. If the palate is cleft, a standard repair is performed at about 6 to 12 months of age, bearing in mind the stability of the airway. If macrostomia is present, this may be corrected at the same procedure. Thereafter, a period of monitoring and orthodontics follows, during which jaw growth is assessed and its function optimized.

Ear reconstruction is performed at approximately 10 years of age, once the child is old enough to understand the procedure, be compliant, and possess sufficient costal cartilage for harvest. Jaw reconstruction may be performed by a variety of methods. Early reconstruction of Pruzansky IIb and III by costochondral rib grafting is an established method, and subsequent growth of the graft makes it suitable for early reconstruction.^{139,140} More recently, distraction osteogenesis has been employed for Pruzansky I and IIa (and above) mandibles and for postreconstructed mandibles.¹⁴¹ Conceptually, this technique is excellent for this reconstructive problem. A caveat, however: its use must be carefully considered in skeletally immature patients in which there is insufficient evidence to support its use because of the high reoperation rate and cost relative to orthognathic surgery at maturity.¹⁴² Although orthognathic surgery at maturity is associated with stable results, there is a higher secondary surgery rate than for other patient groups.¹⁶

Treacher Collins Syndrome (Mandibulofacial Dysostosis)

The previously described syndromes underscore the importance of normal neural crest proliferation, migration, and patterning to the development of the face. The posteriormost stream of the neural crest contributes to the heart (see Fig. 18-1) and links the cardiac defects seen with 22q DS to the underlying developmental process. Although the exact mechanisms remain unknown in 22q DS and OAVS, the neural crest defect in Treacher Collins syndrome has been elucidated.

Treacher Collins syndrome was named after the eminent British ophthalmologist, Edward Treacher Collins, after his description of 11 cases¹⁴³ (11 years after the original description by Berry¹⁴⁴). It is a first branchial arch syndrome that shares many features with OAVS but generally occurs symmetrically. Treacher Collins syndrome affects 1 in 25,000 to 50,000, with an equal sex distribution and no racial predilection. It has an autosomal dominant inheritance with a variable penetrance, but 60% are new mutations.¹⁴⁵ The gene coding for the disorder is *TCOF-1*. The protein product is called *treacle* and is implicated in preribosomal RNA processing; it affects the production of a mature ribosomal RNA for translation into protein. This product is required for neural crest cell proliferation, and haploinsufficiency leads to neural crest apoptosis and consequent hypoplasia of neural crest-derived tissues.^{146,147} Like 22q DS syndrome, it is often regarded as a neurocristopathy.

Clinical Features

The key feature of Treacher Collins syndrome is the so-called *birdlike facies*, in which skeletal hypoplasia in the first arch primarily affects the orbits, mandible and midface, ears, and soft tissues (Fig. 18-16). The similarity to OMENS makes it straightforward for grouping the clinical features.

Orbital Defects Zygomatic hypoplasia with deficiency of the lateral orbital wall leads to a loss of lower eyelid support. The lower lids are hypoplastic with coloboma, absent lashes medially, and downslanting palpebral fissures.

Mandibular and Midface Anomalies The hypoplasia affects the zygoma, mandible, and to a lesser extent the temporal bones. Tessier regarded this as a combined 6, 7, and 8 cleft, with cleft 6 referring to hypoplasia at the zygomaticomaxillary junction, cleft 7 involving hypoplasia of the zygoma with microtia and macrostomia, and cleft 8 being characterized by hypoplasia at



Fig. 18-16 A child with Treacher Collins syndrome, exhibiting severe symmetrical mandibular hypoplasia with ocular coloboma and hypoplastic zygomas, leading to downslanting palpebral apertures.

the frontozygomatic junction. The mandible has a short ramus, body, and prominent antegonial notch, creating a short posterior facial height and a convex facial profile.¹⁴⁸ This causes retrognathia and class II malocclusion, with an anterior open bite. The Pruzansky classification is routinely used for this group.

Ear Defects Patients with Treacher Collins syndrome typically present with symmetrical microtia, with or without atresia of the external auditory meatus, sensorineural deafness, and tongue-shaped sideburns. A hypoplastic middle ear and ossicles is an almost universal finding, and more than 95% have mild to moderate hearing loss as a result of external auditory meatus atresia or ankylosis of the ossicles. A bone-anchored hearing aid (BAHA) is used in these patients.

Nose and Mouth Defects Treacher Collins infants present with choanal atresia or stenosis. In later life the nose may exhibit a broad dorsum with an osteocartilaginous hump and inadequate tip projection. The palate is high and narrow, and more than one third of patients have cleft palate. An additional 30% present with velopharyngeal insufficiency or submucosal cleft palate.¹⁴⁹

Management

As with all craniofacial syndromes, a child with Treacher Collins syndrome should be managed within a multidisciplinary framework that focuses on airway, feeding, vision, and hearing.

Airway The airway is the overwhelming priority: the maxillary hypoplasia constricts the nasal passages, leading to choanal stenosis. Micrognathia with a posteriorly positioned tongue will obstruct the oropharyngeal and hypopharyngeal spaces. Because babies are obligate nasal breathers, and the pharyngeal airway of affected newborns is additionally compromised, urgent airway control soon after birth is essential. A spectrum of airway difficulties may be present and can be managed in the same way as for any craniofacial case.

Feeding Because of airway issues, feeding may be difficult and compounded by a cleft palate. Management, monitoring, and treatment will be as for any child with a cleft palate. Special attention is given to the timing of cleft palate closure, in view of the effect it may have on a compromised airway.

Vision Pediatric neuroophthalmologic assessment is useful to determine acuity, corneal exposure, and extraocular muscle function. If there is corneal exposure, correcting the coloboma is a priority.

Hearing ENT evaluation and formal audiometry, such as the auditory brainstem response test, are essential to plan the fitting of either BAHA or cochlear implants in patients with Treacher Collins syndrome. If one ear has normal function, it is rare to intervene on the contralateral side.

Surgical Management

At birth, airway considerations take priority, and multidisciplinary assessment should occur early to optimize feeding. If the cornea is exposed, conservative measures are instituted until the child is old enough to undergo surgical correction. If the coloboma is small and well managed, repair can be delayed or be combined with palate repair. Most small colobomata can be closed using established eyelid reconstruction principles, with local flap reconstructions used for those involving more than 50% of the eyelid. Between 9 and 12 months in most centers, palate repair is performed in the standard fashion, but this may be delayed for airway considerations. Much of the management of the jaw and ear is similar to that for OAVS; in addition, midface augmentation can be performed to provide lower lid support and improve facial contour. Such midface augmentation can be performed with split calvarial bone grafts or custom implants.¹⁵⁰ Soft tissue suspension, such as a mask lift, can be performed and canthopexies are essential.¹⁵¹ At skeletal maturity, orthognathic surgery is used to improve occlusion, but relapse is common and formal rhinoplasty may be requested to improve appearance.^{148,152}



Fig. 18-17 A child with Nager syndrome. Individuals have severely hypoplastic mandibular features, similar to Treacher Collins syndrome, in concert with radial ray deficiency.

Nager Syndrome

Nager syndrome is characterized by mandibulofacial dysostosis and radial ray deficiency (Fig. 18-17). It is mainly sporadic, but some cases are inherited. It is caused by a heterozygous mutation of the *SF3B4* gene on chromosome 1 in most cases.

Clinical Features

The features of Nager syndrome are similar to those of Treacher Collins syndrome, together with radial ray deficiencies, such as radial aplasia and hypoplasia, radioulnar synostosis, and hypoplastic and triphalangeal thumbs. These children are of low to normal intelligence, and palatal clefting is common, particularly manifested as a hypoplastic palate. Genitourinary abnormalities may also be associated, and other anomalies such as those of the lower limb have been reported but are uncommon. Management is the same as for Treacher Collins patients.

CRANIOSYNOSTOSIS AND CALVARIAL DEVELOPMENT

The developing brain grows at a rapid rate and to accommodate this, the calvarium must expand into postnatal life. This expansion is achieved through two mechanisms: first, the resorption of cranial bone on the dural surface of the calvarium and deposition on the exterior; second, the cranial sutures contribute to osteogenesis by allowing the formation of new bone at the periphery. The sutures close postnatally in a coordinated manner after the brain has achieved its full size. Craniosynostosis is the condition in which premature fusion of a suture occurs, restricting growth between the fused bones and reorienting this growth in an abnormal manner, which results in an abnormal head shape. The shape depends on which suture (or sutures) are fused (see Chapter 23).

The craniosynostoses can be classified as nonsyndromic and syndromic, with nonsyndromic presenting as isolated premature suture fusions, and syndromic being more complex, typically presenting with multiple suture fusions and/or other anomalies. The classic syndromic cranio-

synostoses include Apert syndrome, Crouzon syndrome, and Pfeiffer syndrome. Because they also affect the cranial base, they will be considered in the section on cranial base.

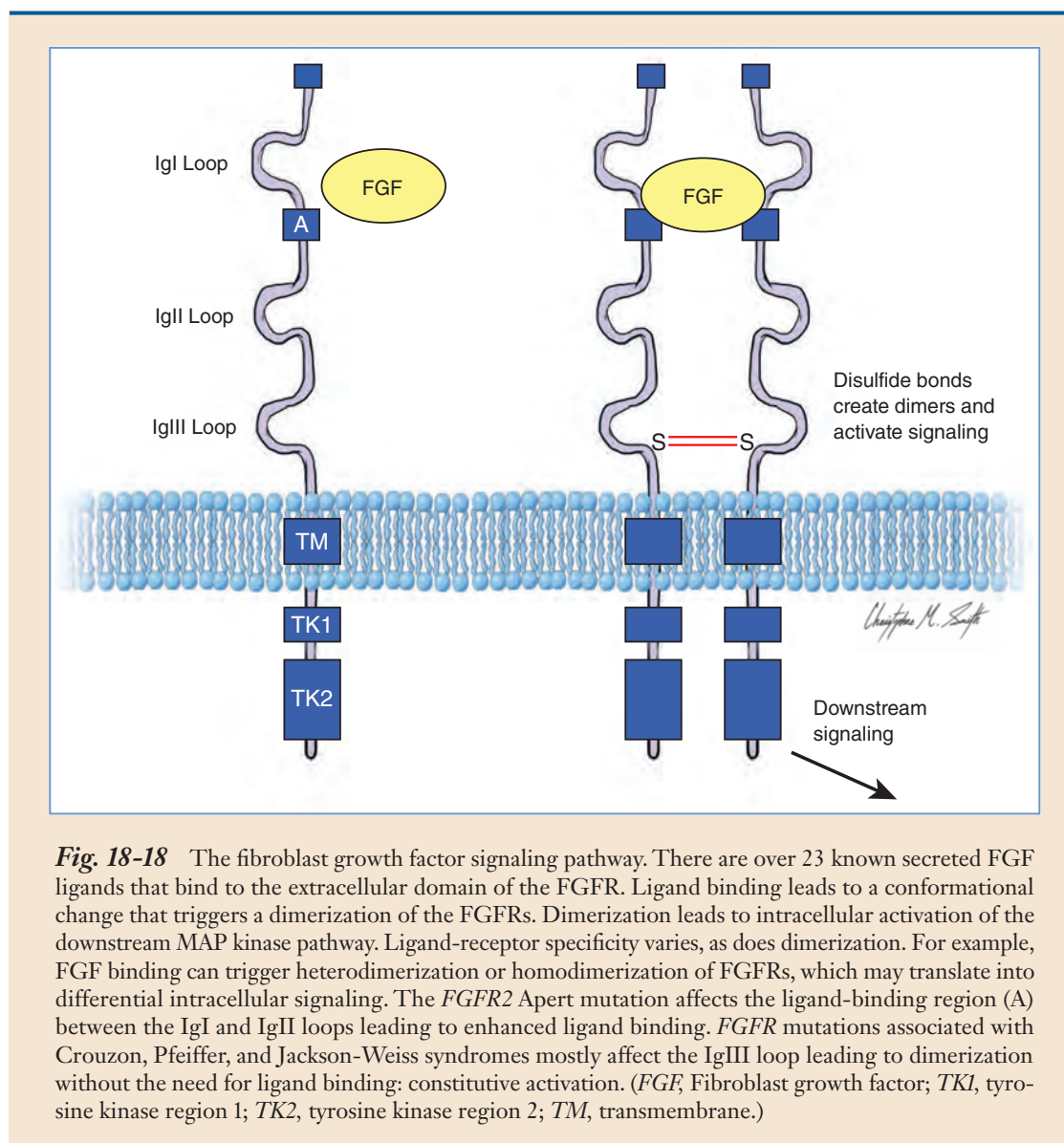
Developmental Anatomy

The development of the skull begins at about 25 days' gestation, originating from both the neural crest and the cranial mesoderm. The skull comprises the neurocranium, which houses the brain, and the viscerocranium, which is derived from the branchial arches and forms the face. The neurocranium consists of the cranial base, also called the *basicranium* or *chondrocranium* (referring to its endochondral origin), and the calvarium that is formed by ossification in membrane and creates the frontal, parietal, squamous, temporal, and part of the occipital bones. The maintenance of sutures is dependent on the presence of the dura.¹⁵³ Initially thought to have a mechanical role, the dura is the source of numerous signaling molecules that are involved in coordinating cranial morphogenesis.

Centers of ossification are seen for the paired frontal and parietal bones at 8 weeks' gestation, and by 14 weeks there is widespread ossification of all bones of the calvarium. Ossification begins by condensation of the mesenchyme cells of the skeletogenic membrane. There may be single or multiple closely spaced centers of ossification for each bone, and these coalesce and spread outward; the advancing edge is called the *osteogenic front*. Ossification expands radially until two areas of ossification confront each other, creating a suture. The sutures are simple adaptive ankyloses between the neighboring calvarial bones; these allow the brain to expand to reach its adult dimensions by growth perpendicular to the orientation of the suture. Mapping studies using transgenic mice have demonstrated that the coronal and sagittal sutures represent a boundary between the neural crest and mesoderm-derived calvarium.¹⁵⁴

Signaling Mechanisms in Suture Development

Once a suture has formed, it needs to remain patent to accommodate brain growth and lay down new bone at the osteogenic front. As elsewhere in the body, conserved signaling mechanisms govern the process of ossification; syndromic craniosynostoses have been found to be caused by mutations in *FGFR* genes among others, and the expression of these molecules has provided clues to the mechanism of suture formation and closure¹⁵ (Fig. 18-18). *Fgf9* is expressed at high levels in the suture mesenchyme, underlying dura and overlying skin.¹⁵⁵ *Fgf2* is expressed in the sutural mesenchyme and its receptors, *Fgfr1* and *Fgfr2*, in the osteogenic fronts indicating a paracrine function. FGF2 is known to stimulate osteoblast proliferation and inhibit differentiation,¹⁵⁶ whereas application of FGF4 on the osteogenic front accelerates suture closure and mimics the FGFR syndromes.¹⁵⁵ *Fgfr1* is expressed in differentiating osteoblasts and *Fgfr2* is expressed in the proliferating osteogenic precursors. *Bmp2* and *Bmp4* are also expressed in the osteogenic fronts, with levels falling after birth: FGF signaling activates *Msx1* and BMP activates both *Msx1* and *Msx2*, with both being expressed in the suture with *Msx2* levels dropping after birth. *Msx2* has a role in maintaining suture patency, and mutations in *MSX2* have been identified in Boston-type craniosynostosis.¹⁵⁷ Signaling networks appear to change after birth with SHH/*Ptc* expression appearing in the osteogenic front at the end of embryonic development. SHH may interact with *Bmps* through a *Ptc*-dependent pathway to maintain suture patency postnatally.¹⁵⁵ The secreted BMP antagonist *Noggin* is expressed postnatally in the suture mesenchyme of patent but not fusing sutures. *Noggin* prevents suture fusion in vitro and in vivo, and it has been hypothesized that



premature *Noggin* downregulation may lead to craniosynostosis; however, no *NOGGIN* (or *BMP*) mutations have been identified in human cases.¹⁵⁸

Although much remains to be understood regarding suture growth and fusion, the role of the FGF and related pathways is beginning to emerge from a combination of transgenic mice and in vitro assays. One model suggests that FGFs are secreted by osteoblasts and diffuse toward the osteogenic fronts¹⁵⁹ (Fig. 18-19). At low concentrations, the FGF stimulates proliferation of sutural stem cells and *Fgfr2* expression. As the bone matrix advances toward the osteogenic front, the FGF concentration rises and the proliferating cells initiate differentiation into *preosteoblasts*. This is accompanied by downregulation of *Fgfr2* and upregulation of *Fgfr1* expression, whereupon they begin to secrete matrix and are called *osteoblasts*.¹⁵⁹ Exogenous application of FGF2 upregulates *Twist* expression in the sutural mesenchyme. *Twist*'s role is to inhibit the terminal differentiation of osteoprogenitor cells into osteoblasts. FGF2 has stimulatory and inhibitory effects, and in-

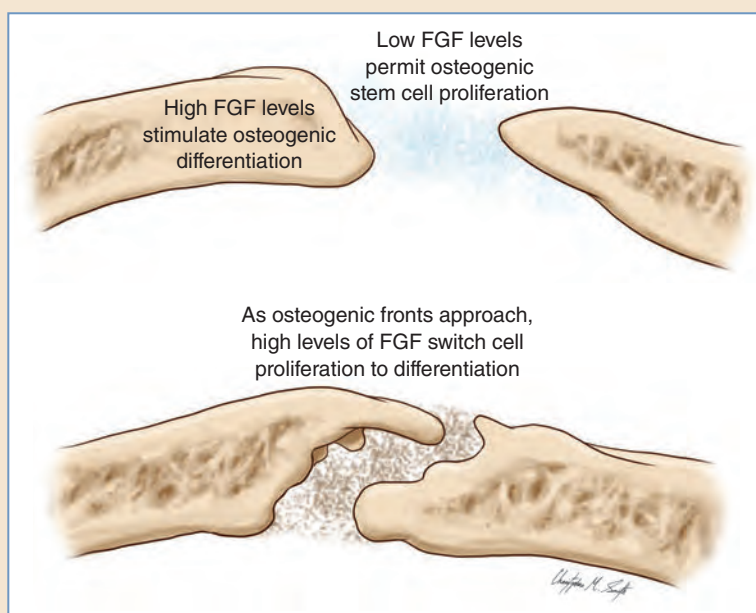


Fig. 18-19 FGF control of proliferation and differentiation at the calvarial suture osteogenic front. A proposed model is based on a balance of FGFR1 and FGFR2 expression regulated by the concentration of FGF ligand. (FGF, Fibroblast growth factor.)

hibitory effects appear to be mediated in a *Twist*-dependent fashion.¹⁵⁶ Loss of function of twist disinhibits osteoblast differentiation, leading to premature closure of the suture, and this loss of function has been identified as the cause of Saethre-Chotzen syndrome.^{160,161} It appears that the quantity of TCF12-Twist heterodimers is critical in coronal suture development. *TCF12* haploinsufficiency causes coronal synostosis in humans, and haploinsufficiency of both *Tcf12* and *Twist* in mice leads to a severe bilateral coronal synostosis.¹⁶² For example, *Fgfr3* exhibits weak expression in skeletogenic membrane and stronger expression in skull base cartilage. However, mutations in *FGFR3* cause Muenke syndrome and may reflect a coronal suture-specific function of *FGFR3*.¹⁶³

DEVELOPMENT OF THE CRANIAL BASE AND FACIAL DYSOSTOSES

The morphologically complex cranial base suspends the face and ossifies from cartilage (chondrocranium). The chondrocranium consists (from rostral to caudal) of the paired trabeculae cranii that lie between the paired nasal capsules, the *sphenoid* (comprising paired orbitosphenoid and alisphenoid cartilages), and the basioccipital cartilage derived from the paired parachordal cartilages (either side of the notochord). A detailed account of the developmental anatomy of the cranial base has been published elsewhere.¹⁵

The chondrocranium is completed by 12 weeks' gestation, and primary ossification centers emerge within this unified cartilaginous structure¹⁶⁴ (Fig. 18-20). The synchondroses are the joints that remain between the bone centers. The sphenoethmoidal fuses at 6 years and the intrasphenoid ossifies at birth. By contrast, the sphenooccipital synchondrosis plays an important role in postnatal growth and fuses around puberty.¹⁶⁵ Synchondroses can be considered a mirror image of an epiphyseal growth plate of a long bone in terms of morphology and possibly chemical

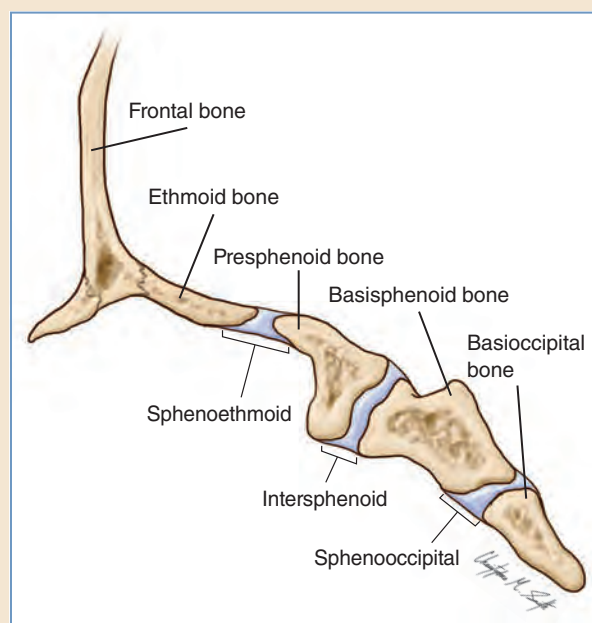


Fig. 18-20 The cranial base and synchondroses.

signaling. They comprise proliferating and hypertrophic zones flanking a central reserve zone. By this arrangement, they produce growth in either direction, with a net effect of lengthening the cranial base.

In the growth plates of the appendicular skeleton, *Indian hedgehog* has a critical role in governing bone growth, acting as a paracrine factor that stimulates chondrocyte proliferation and inhibiting maturation. *Ihh*^{-/-} mice have shorter cranial bases, with a disorganized histologic appearance correlated to lower chondrocyte proliferation, and low levels of *Ptc* and *PTHrP*.¹⁶⁶ This is more marked in the rostral synchondroses. More posteriorly, SHH may compensate, and it is strongly expressed by notochord remnants adjacent to sphenooccipital synchondrosis but not in the more rostral intrasphenoidal synchondrosis. Examination of the role of FGFR3 signaling in an achondroplasia model (gain of function FGFR3) demonstrated upregulation of BMP with reduced chondrocyte proliferation and early ossification of the synchondrosis.¹⁶⁷ BMP4 has been shown to upregulate hedgehog signaling and increase chondrocyte proliferation and endochondral ossification in a dose-dependent manner.¹⁶⁸ Other signaling pathways are also involved, including the Wnt pathway. By knocking out its downstream mediator β -catenin, the synchondroses demonstrated mainly resting chondrocytes without growth plate organization. Conversely, constitutively active β -catenin led to precocious chondrocyte maturation and hypertrophy in a topographically disorganized fashion. A balance between *Ihh* and Wnt signaling may play a role in the balance between chondrocyte proliferation and maturation.¹⁶⁹

Carpenter Syndrome

Carpenter syndrome is rare, with fewer than 100 cases reported in the literature to date. Unlike other acrocephalosyndactylies, it is autosomal recessive but often presents as new mutations with great phenotypic variability.¹⁷⁰ Mutations in *RAB23*, a negative regulator of hedgehog sig-



Fig. 18-21 Carpenter syndrome is rare. Kleeblattschädel deformity (cloverleaf skull) is characteristic of the syndrome and refers to the head shape as a consequence of multisuture synostosis.

naling, have been identified, linking the laboratory knowledge of *hedgehog* and suture biology to the clinic.¹⁷¹

Clinical Features

Carpenter syndrome is characterized by craniosynostosis, preaxial polydactyly of the feet, brachydactyly, syndactyly, obesity, and developmental delay (Fig. 18-21). Synostosis typically begins at the sagittal suture, then to the lambdoid and coronal sutures in succession. The anterior cranial fossa is underdeveloped, with shallow orbits that have an unusually “tall” morphology. Patients may exhibit hypotelorism or hypertelorism, and the eyes may feature ptosis; corneal opacity and optic atrophy may occur. There is no correlation between cranial dysmorphology and neurologic abnormality.¹⁷² Ears are low-set and may be rotated posteriorly; preauricular sinuses and fistulas may occur. Developmental delay is usual, but cases of normal intellect have been reported.¹⁷³ Other anomalies, such as umbilical hernia/omphalocele, short stature, hypogonadism and obesity, have all been reported.

The FGFR and Related Craniosynostosis Syndromes

Apert syndrome has a relatively narrow phenotype. By contrast, other syndromes are much more heterogeneous in their phenotype and overlap clinically. Genetically, the syndromes of this group have multiple different mutations and will be considered together. FGFRs have multiple roles during ossification through regulation of chondrocyte and osteoblast proliferation and differentiation as well as differing effects at each of the various cranial sutures. By acting on multiple processes during suture/synchondrosis formation, great clinical heterogeneity is observed.

Apert Syndrome

Originally described by Wheaton in 1894,¹⁷⁴ Apert syndrome is known eponymously for French physician Eugène Apert, who described a series of cases. It is the second most common facial dysostosis syndrome after Crouzon syndrome. The incidence is 1:160,000 live births, with most cases being sporadic, but autosomal dominance has been reported.



Fig. 18-22 Apert syndrome. **A** and **B**, Kleeblattschädel deformity (cloverleaf skull). The lateral view shows ventriculoperitoneal shunts in place. **C** and **D**, A 6-month-old infant with Apert syndrome and airway compromise requiring early tracheostomy.

Phenotype

The phenotypic features of Apert syndrome can be grouped into craniosynostosis with intracranial pressure problems, midfacial hypoplasia, symmetrical syndactyly of the hands and feet, and other features (Fig. 18-22).

Craniosynostosis and Intracranial Pressure Children with Apert syndrome typically present with a brachycephalic head shape as a result of bicoronal synostosis and a large anterior fontanel that forms a bregmatic bump (volcano sign), because the increased pressure pushes against areas of weakness in the vault. It may also be associated with multiple suture synostosis and cloverleaf skull, in which case mental retardation can be profound and the prognosis is poor. The anterior fontanel remains patent for a prolonged period if it is left untreated.

Raised intracranial pressure occurs in 43% of Apert syndrome patients, but learning difficulties are more common, likely because of other brain anomalies, such as hydrocephalus, agenesis of the corpus callosum, hypoplastic white matter, and ectopic gray matter. Developmental delay

is mostly mild, and the individual may attain a high level of mental function.¹⁷⁵ Raised intracranial pressure in syndromic synostoses is not purely a matter of cerebrocranial disproportion; in fact, the Apert cranial volume is greater than in the normal population.¹⁷⁶ A complex interplay of other factors contributes to intracranial hypertension, including obstructive hydrocephalus, anomalous venous drainage that creates venous hypertension with “back pressure” on the CSF, and obstructive sleep apnea that leads to increased CO₂ and therefore cerebral venous dilation.¹⁷⁷

Midfacial Hypoplasia Premature fusion of the sphenoccipital, sphenofrontal, sphenovomerine, and vomeromaxillary sutures in children with Apert syndrome leads to midfacial hypoplasia, with specific implications for the jaws and orbits.^{178,179}

Exorbitism, a protrusion of the globe as a result of reduced orbital volume, is caused by verticalization of the orbital roof and the hypoplastic inferior orbital rim. The ethmoids are “prolapsed” inferiorly between the orbits, bringing the cribriform plate almost as low as the equator of the globe and possibly accounting for the orbital hypertelorism and divergent visual axes.¹⁸⁰ As a result, the globe is situated anteriorly and the eyelids may be incompetent, leading to corneal exposure; this has been called *malignant exorbitism* and mandates early ocular protection. Other mechanisms contribute to visual dysfunction, such as optic nerve atrophy (papilledema) resulting from raised intracranial pressure. Extraocular muscle abnormalities may also contribute to disorders of conjugate gaze. *V-pattern exotropia* (divergent upgaze and esotropic downgaze) is more common in Apert syndrome than in Crouzon syndrome, possibly because of the absence of the superior rectus and architectural abnormalities of the extraocular muscles.¹⁸¹ This affects the maxilla to a greater degree than the zygoma and is a function of the reduced growth of the skull base; as a result, the pterygomaxillary fissure is virtually obliterated. An anterior open bite with premature posterior contact occurs, and the alveolar arch faithfully reflects the maxillary hypoplasia with a narrow crevicolike morphology to the hard palate; 75% of patients have a cleft palate with variable expressivity.¹⁸² This results in a crowded pharynx and a narrow nasal airway and may cause severe respiratory distress, especially during the period of obligate nasal respiration. The necessity of airway interventions is common in infants, whereas obstructive sleep apnea is frequent in older patients, and sleep studies are often required for monitoring. Almost all patients have otitis media, with effusion that persists into adult life, with more than half developing a permanent conductive hearing loss.¹⁸³

Hand Anomalies The asymmetrical syndactyly of the hands and feet seen in children with Apert syndrome is detailed in Chapter 45. The key features are complex or complicated syndactyly of the middle three rays where the tips (Greek *acros*) may be joined, a shortened radially deviated thumb, simple syndactyly of the fourth web, and brachyphalangism.

Other Features Other features of Apert syndrome are vertebral fusions, a “beaked” nose, cardiopulmonary abnormalities, and hyperseborrhea manifested as moderate to severe acne with unusual extension to the forearms. Drooling is also an issue, and the tracheal cartilages may form plates or a sleeve rather than rings, making intubation for procedures complex.

Genotype

Apert syndrome is autosomal dominant, with many cases being new mutations. It has a relatively narrow genotypic spectrum, with 98% of cases caused by one of two mutations in the FGFR2 IgI-IgII linker region, which leads to increased affinity and reduced specificity for the FGFR ligand, encouraging dimerization and increasing FGF signaling.¹⁸⁴ *FGFR2^{P253R}* constitutes 37% of identified mutations and is associated with a more severe limb phenotype; *FGFR2^{S252W}* is associated with cleft palate and constitutes the remaining 63%.¹⁸⁵

Developmental Implications A postmortem examination of one Apert syndrome patient showed premature fusion of the sphenoccipital synchondrosis and fusion of the vomer to the sphenoid and maxilla.¹⁷⁸ *FGFR2* is primarily expressed in midfacial membranous ossification, and

it has been shown that *FGFR2* gain-of-function promotes an increased number of cells to enter osteoblastic differentiation, leading to premature ossification.¹⁸⁶ The transgenic mouse model of Apert syndrome (*FGFR2*^{+/*S252W*}) demonstrated that individual cranial sutures exhibited different histologic changes; for example, the “metopic” suture exhibited increased proliferation and delayed osteoblastic differentiation, whereas in the sagittal suture osteogenic markers were increased. The basicranium had increased cartilage, and ectopic cartilage was seen in some organs, including the trachea and between calvarial bones.¹⁸⁷ This indicates that defects in chondrogenesis also play a role in Apert syndrome.

Much of the evidence suggests that Apert syndrome is a gain of *FGFR2* function that leads to an abnormal chondrocranial template, with premature ossification reducing rostrocaudal growth contributing to the midfacial hypoplasia.^{164,186,187} Membranous ossification of the calvarium is affected through abnormal osteoblastic proliferation and differentiation.

Crouzon Syndrome

Crouzon syndrome is the most common of the syndromic craniosynostoses. It is typically described as being similar to but less severe than Apert syndrome in appearance, but the craniofacial phenotype is different at all ages.¹⁸⁸ It constitutes 5% of all craniosynostoses, with a prevalence of 16.5 per 1 million¹⁸⁹ (Fig. 18-23).

Phenotype

Craniosynostosis With Chiari Malformation A variable pattern of craniosynostosis can occur in Crouzon syndrome patients, ranging from mild cases with little sutural involvement to more severe cases with multiple synostosis of a severe and progressive nature, including the cloverleaf skull.¹⁹⁰ In contrast to the skull of a patient with Apert syndrome, where the bregma synostoses by the formation of bony islands, a patient with Crouzon syndrome exhibits bicoronal synostosis that may progress to other sutures. The skull base synchondroses were found to be fused earlier than in Apert syndrome, and this together with the multiple craniosynostosis was suggested to

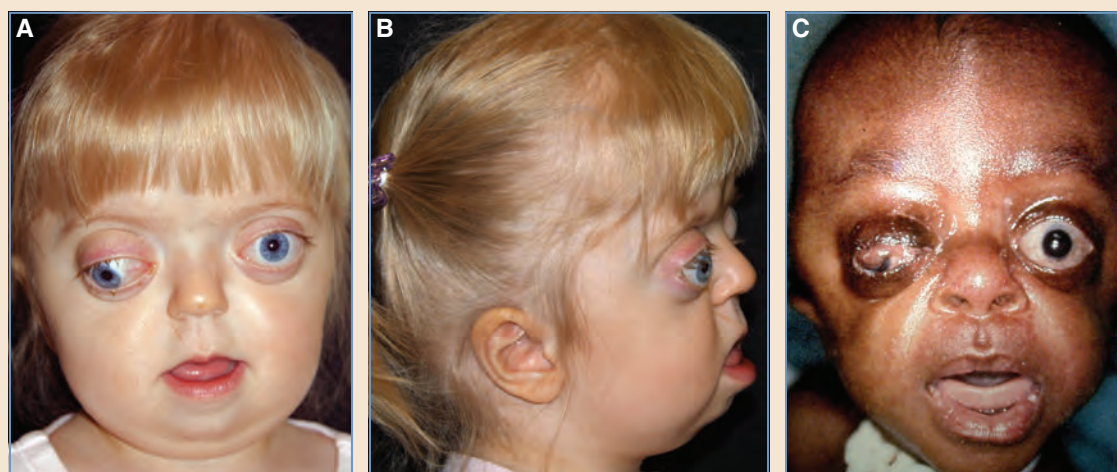


Fig. 18-23 Crouzon syndrome. **A** and **B**, Frontal and lateral views of an affected girl. **C**, Another patient with severe exorbitism, placing the globes at risk.

account for the increased intracranial pressure.¹⁹¹ Developmental delay is unusual, but headaches, seizures, and hydrocephalus are known to occur. Chiari malformation is found in up to 70% of Crouzon syndrome patients. It is postulated that fusion of the lambdoid suture together with cranial base synostosis produces alterations in the skull base, creating a small posterior fossa with herniation of the cerebellum into the cervical canal.¹⁹²

Exorbitism The lateral and inferior orbital rims are markedly hypoplastic, with the globe in a normal position relative to the cranial base. This is in contrast to the orbit of an Apert syndrome patient, in which the posterior orbital wall protrudes the globe.¹⁸¹ Optic atrophy is found in 20% of patients with Crouzon syndrome but is not characteristic of Apert syndrome patients.

Midfacial Hypoplasia Midfacial hypoplasia in Crouzon syndrome patients is less severe than in Apert syndrome patients but may have the same issues: respiratory compromise and obstructive sleep apnea resulting from the pharyngeal hypoplasia. One third of children with Crouzon syndrome are obligate mouth breathers because of their small nasopharynx. Cleft palate is rare, although a narrow palate occurs. There is dental crowding and a constricted arch as a consequence of maxillary hypoplasia with class III occlusion.

Other Features Other features include fusions between the C2-3 and C5-6 vertebrae, which occurs in up to 30% of Crouzon syndrome patients.¹⁹³ Hearing loss is common, with approximately one half of patients having conductive hearing loss.

Genotype

Crouzon syndrome is associated with *FGFR2*^{C342R} and *FGFR2*^{C342Y} mutations that lead to an unpaired cysteine residue, allowing ligand-independent cross-linking of the receptor dimers and constitutive activation.¹⁹⁴ These mutations have also been shown to cause Pfeiffer syndrome and Jackson-Weiss syndrome.¹⁹⁵

Pfeiffer Syndrome

Phenotype

Pfeiffer syndrome is an autosomal dominant disorder with incomplete penetrance and variable expressivity; it affects 1 in 100,000 live births.¹⁹⁶ The phenotypic spectrum is similar to that of Apert and Crouzon syndromes: a short anterior cranial base, midfacial hypoplasia with a class III occlusal relationship, an anterior open bite, a narrow high-arched palate, low-set ears, and conductive hearing loss. Pfeiffer syndrome is characterized by broad, medially deviated toes and thumbs, and radiohumeral synostosis with flexion deformity of the elbow. Cervical fusions, among other skeletal anomalies, have been reported.

Pfeiffer syndrome has been classified into three types by Cohen¹⁹⁷:

Type 1 Classic Pfeiffer: Isolated symmetrical coronal synostosis, broad thumbs and great toes, and incomplete syndactyly of the second web

Type 2 Cloverleaf skull: Severe midfacial hypoplasia and exorbitism, intracranial involvement with developmental delay, and ankylosis of the elbows

Type 3 Severe: Type 2, without the cloverleaf skull

Like the other facial dysostoses, Pfeiffer syndrome exhibits a brachycephalic calvarial morphology, and multisuture synostosis may cause more severe morphologies, including malignant exorbitism, midfacial hypoplasia, and airway issues.¹⁹⁸ Chiari malformation occurs in 50% of children with Pfeiffer syndrome, increasing to 100% in those with cloverleaf skull.¹⁹² Types 2 and 3 have a poor prognosis, with developmental delay and a limited lifespan.^{197,198} It has been suggested that a proactive approach with early tarsorrhaphies, aggressive airway management, and frequent screening for Chiari malformation may reduce mortality¹⁹⁹ (Fig. 18-24).

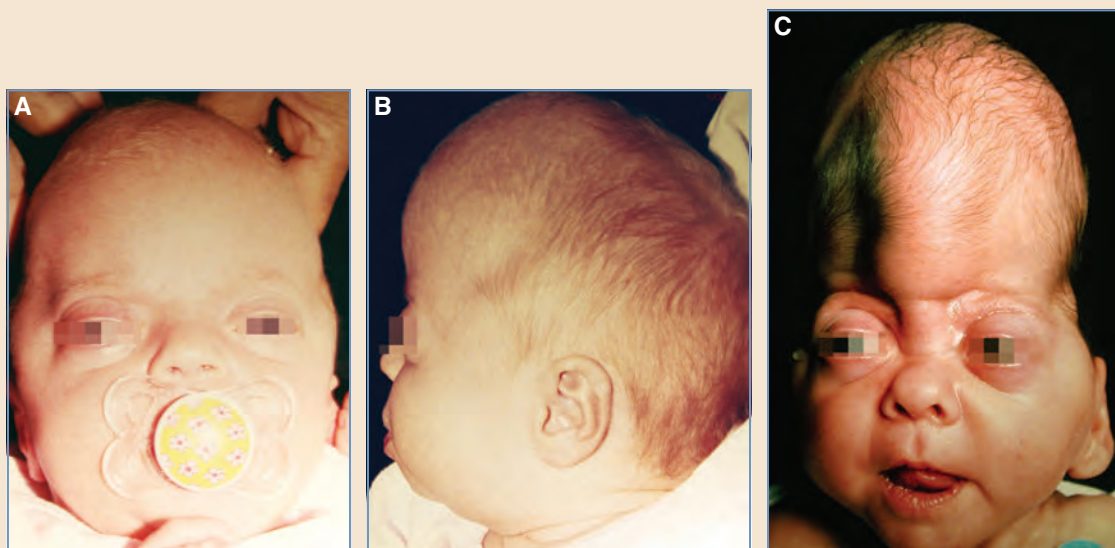


Fig. 18-24 Pfeiffer syndrome. **A** and **B**, This infant has type 1 Pfeiffer deformity with bilateral coronal synostosis. **C**, This infant has a type 2 deformity with pansynostosis and Kleeblattschädel deformity.

Genotype

The majority of patients with Pfeiffer syndrome have a mutation in *FGFR2*, but approximately 5% have a mutation in *FGFR1*.²⁰⁰ *FGFR1*^{P252R} results in the milder type 1 form of Pfeiffer syndrome, whereas types 2 and 3 are caused by *FGFR2* mutations only and may have abdominal anomalies such as prune belly syndrome and malrotations.²⁰¹ *FGFR2*^{C342Y} and *FGFR2*^{C342R} are responsible for cases of Crouzon and Pfeiffer syndromes, and *FGFR2*^{C342R} also results in Jackson-Weiss syndrome.²⁰² These mutations affect the IgIII loop and cause it to unfurl, allowing it to form heterodimers/homodimers and activate downstream signaling, irrespective of ligand activation. The reasons for the phenotypic differences in Pfeiffer syndrome patients remain to be explained but are likely to include modifying genes, variable ligand receptor activation, and epigenetic factors.

Jackson-Weiss Syndrome

Jackson-Weiss syndrome is a clinically distinct facial dysostosis characterized by bony anomalies of the feet, including a broad, short metatarsal and proximal phalanx of the great toe, abnormal tarsal bones, and tarsal/metatarsal coalitions. Involvement of the hands is generally not observed, but the similarity to Pfeiffer syndrome has been highlighted.¹⁹⁷

Developmental Implications

The underlying mechanism of Jackson-Weiss syndrome is the premature synostosis of the skull base and calvarial sutures. Clues to the mechanism come from the *FGFR2*^{C342Y} mouse, with a mutation equivalent to Crouzon and Pfeiffer syndromes.³ A child with Jackson-Weiss syndrome exhibits Crouzon features with invariant fusion of the coronal and partial fusion of the sagittal and lambdoid sutures. There is also progressive fusion of the synchondroses of the cranial base.²⁰³ Results suggest that *FGFR2* regulates the osteoblast proliferation and differentiation, resulting in enhanced osteogenesis. In the calvaria, more osteoprogenitor cell proliferation is observed. By contrast, the synchondroses show lower than normal chondrocyte proliferation; this is suggested to occur because the interaction of *FGFR2* with *FGFR3* during endochondral ossification demonstrates the different effects of the mutation on chondrocyte and osteoblast differentiation.

Muenke Syndrome

Nonsyndromic craniosynostosis was thought to be a sporadic phenomenon, but studies showed that approximately 30% of unicoronal synostosis had a genetic component.²⁰⁴ The most common of these was identified as an *FGFR3*^{P250R} mutation called *Muenke syndrome*, which affects approximately 10% of nonsyndromic unicoronal cases and 50% of nonsyndromic bicoronal cases.²⁰⁴ The equivalent mutation in *FGFR1* causes Pfeiffer syndrome and in *FGFR2* causes Apert syndrome.

There is significant phenotypic variability in patients with Muenke syndrome; however, *FGFR3*^{P250R} phenotypes lack the midfacial hypoplasia and have a variable craniosynostosis and limb involvement. They may have severe coronal synostosis (unilateral or bilateral), but the morphologic severity does not correlate with raised intracranial pressure.²⁰⁵ Learning difficulty in this group remains to be conclusively defined but may affect a minority of patients. In one series, all patients with Muenke syndrome had bilateral mild low- to mid-frequency sensorineural hearing loss and a 44% revision rate of frontoorbital remodeling as a result of recurrent deformity.²⁰⁴

Saethre-Chotzen Syndrome

Saethre-Chotzen syndrome is a heterogeneous autosomal dominant syndrome with variable expression, typically presenting with variable coronal synostosis, prominent crus helices, low frontal hairline, blepharoptosis, and incomplete simple syndactyly of the second web space. However, the presentation may include multisuture synostosis or no craniosynostosis, other limb anomalies, and ophthalmic issues such as lacrimal duct stenosis, strabismus, and pseudohypertelorism.²⁰⁶ Learning difficulties are uncommon in children with Saethre-Chotzen syndrome.²⁰⁷ The majority of patients have a mild dysmorphism; those with craniosynostosis are typically managed by frontoorbital remodeling, but the reoperation rates are comparable to those for Muenke *FGFR3* synostosis.

Although most patients with Saethre-Chotzen syndrome have a *TWIST* mutation, some patients with an overlapping phenotype have a mutation of *FGFR2* or *FGFR3*.²⁰⁷ The variability in the *TWIST1* mutation (more than 100 mutations) may account for the variable phenotype, but the nature of the mutation has not been shown to correlate with the severity of the craniosynostosis or the need for secondary surgery²⁰⁶ (Fig. 18-25).



Fig. 18-25 This 3-year-old child is one of four siblings who showed variable expression of Saethre-Chotzen syndrome inherited as an autosomal dominant trait from their affected father.

FGFR SYNDROME GENOTYPE-PHENOTYPE CORRELATIONS

We have discussed a family of related craniofacial dysplasias with similar phenotypes and a common underlying genetic origin. Nonetheless, a simple mutation-phenotype relationship does not exist; for example, the same mutation can lead to both Crouzon and Pfeiffer syndromes, and multiple mutations can cause the same single eponymous syndrome.^{200,202} This diversity arises from the nature of the FGF signaling pathway, which has great variability at a number of steps. Once the FGFR signaling system is established, growth depends on the balance between the proliferation of chondroprogenitor and osteoprogenitor cells with bone and cartilage maturation. Imbalances at different steps of the pathway lead to disparate phenotypic changes.

Whereas 98% of Apert syndrome cases are caused by two mutations in the *FGFR2* IgI-II linker region that confer increased ligand binding, Pfeiffer and Crouzon syndromes and related phenotypes show greater phenotypic heterogeneity and primarily arise from a wide range of mutations in the *FGFR2IIIc* region that creates a ligand-independent activation of the receptors. Pfeiffer syndrome may allow more promiscuous ligand binding but “normal” receptor dimerization and signaling, with a tighter phenotypic spectrum. Crouzon syndrome is ligand independent; the mutations may not only homodimerize but also heterodimerize, with FGFR1 and FGFR3 likely resulting in differing degrees of downstream signaling and a variable degree of ossification. Therefore IgIII mutations lead to heterodimerization, promoting cranial osteogenesis beyond the negatively autoregulated FGFR2 domain.²⁰⁷ By doing so, the regions of the skull that express *FGFR1* and *FGFR3* may become recruited into the pathologic process, leading to multisuture synostosis.¹⁶⁴

Complicating this are the multiple splice variants of the IgIII loop that exhibit differential ligand binding and tissue specific expression. Superimposing all of this onto different genetic backgrounds and environmental factors explains why it is so hard to draw simple associations between mutations and phenotypes.

PRINCIPLES OF MANAGEMENT OF SYNDROMIC CRANIOFACIAL PATIENTS

All craniofacial patients should be managed within a multidisciplinary framework that includes a craniofacial surgeon, otorhinolaryngologist, ophthalmologist, orthodontic/pediatric dentist, speech and language therapist, dietician, psychologist, and pediatrician. The issues faced by individuals with different syndromes can be categorized systematically, because the management for each is similar, irrespective of the syndrome.

Airway Considerations

Airway issues for a syndromic craniofacial patient may be potentially catastrophic when presenting at delivery, or mild through severe when presenting soon after birth. Emergency airway management in the delivery suite is not considered here; subsequent management is important to facilitate feeding and prevent further complications.

In a neonate with respiratory difficulty, an initial task is to nurse the child in a side-to-side or prone position; nursing the child in a prone position allows a large tongue to sit forward in the mouth, clearing the hypopharynx. A large proportion of babies respond well to such conservative measures, and their airway control improves as they gain weight. If positioning does not improve the airway, then a nasopharyngeal airway (NPA) should be considered. An NPA is indicated if the obstruction is not alleviated by positioning, if the child's airway is obstructed when he or she is handled, or if there is increased airway difficulty with feeding. The NPA may be left in situ for a significant length of time, but if such an intervention is required long term, a tracheostomy should be considered. Before this, a thorough endoscopic evaluation of the airway should be performed to exclude multilevel obstruction, such as laryngomalacia, tracheomalacia, and bronchial stenosis; the level of obstruction will guide the intervention. Additionally, central causes, including hypotonia and central apneas, should be considered, and sleep studies may be of value. Choanal atresia is usually apparent early on, because neonates are obligate nasal breathers. The otolaryngologist plays a key role in evaluation and performing dilation (often repeated) or stenting of the obstruction. The hypoplastic nasopharynx in the facial dysostoses may be expanded by a LeFort III or monobloc osteotomy, but this is usually reserved for children with additional malignant exorbitism and raised intracranial pressure. Such procedures in very young children are associated with significant risk.

Feeding Considerations

A feeding assessment by an experienced practitioner is invaluable. One should be alert to signs of obstruction to an otherwise stable airway when feeding, such as slow feeding, falling asleep during feeding, and failure to gain weight. Conservative measures in cleft palate babies include the use of squeezable MAM bottles or Haberman feeders, with which the caregiver delivers the milk to the back of the throat. Continued poor intake mandates a nasogastric tube; the child may be re-evaluated periodically to try to return to oral intake. If long-term nasogastric feeding is required, consideration should be given to a feeding gastrostomy; this is more common in children with associated neurologic abnormalities.

Vision Considerations

Exposure of the globe can lead to blindness from corneal ulceration and even perforation. Impending exposure can be managed as for facial palsy patients, with lubrication using drops by day and ointment at night.^{208,209} If the limbus is observed during sleep and associated with ocular surface symptoms in the morning, then taping the eyelid shut at night is recommended. If this is not tolerated, consideration should be given to a temporary tarsorrhaphy; this is especially important in malignant exorbitism or repeated globe subluxations. The family can be shown how to reduce the globe after subluxation.

Visual assessment by an ophthalmologist is recommended in virtually all syndromic patients to identify further anomalies, institute early management of such conditions as dysconjugate gaze, and monitor for papilledema. Some centers use visually evoked potentials to monitor for increased intracranial pressure.

Hearing Considerations

In most countries a hearing test is available for newborns; suspicion of hearing issues should prompt further assessment. Auditory brainstem potentials can be performed with the infant under anesthetic in concert with another procedure; this gives information regarding sensorineural hearing. An ENT examination is important to identify and treat glue ear. Hearing tests in young children can be challenging, but they provide valuable information once a baseline is achieved. Provision of hearing aids is important for speech development; however, in instances of microtia, the decision to use a BAHA should be made as a team to avoid limitation of potential reconstructive options.

Speech Considerations

Speech therapists are vital members of the craniofacial team, performing regular speech assessments and therapy and providing advice to families as required. Typically an informal speech assessment is performed at 18 months of age, with a formal assessment at 3 years. Given the propensity for cleft palate in craniofacial patients, most of the management is focused on identifying and managing velopharyngeal insufficiency.

Psychology and Patient-Reported Outcome Measures

Facial difference has a huge impact on the patient and the family. The psychologist aims to explore these issues, providing advice and coping strategies tailored to the age and circumstances of the patient and family. Apart from surgical correction for functional reasons, much surgery is focused on “normalizing” the facial difference. It has become clear that the opinion of the patient and family regarding the quality of outcomes of treatment is important in guiding interventions. Recently, patient-reported outcome measures for facial aesthetics and cleft patients have been introduced. Such instruments are expected to become widespread in the future.

CONCLUSION

The combination of developmental biology and genetics heralds a new era for the diagnosis and classification of craniofacial differences. This chapter should equip the surgeon with a sufficient understanding of craniofacial development to understand the pathologic anatomy that will be encountered in the clinic. A systematic approach for the assessment and management of the child with craniofacial disorders is presented.

KEY POINTS

- Genes code for proteins, not anatomic structures or syndromes. Many genes have overlapping functions. Their action is modified by other genes and/or environmental factors: epigenetics.
- Genetic models exist for many human syndromes and provide insight into the mechanisms of disease.
- Craniofacial development is a stepwise process that consists of creating and then refining the morphology of the embryo.
- Midline facial development is intimately linked to brain development and underlies a spectrum of midfacial defects from holoprosencephaly to fronto-nasal dysplasias.
- Abnormal fusion of the facial processes is the cause of many typical and atypical facial clefts.
- Cleft palate may be a result of a failure of palatal growth, elevation, or fusion.
- Abnormal neural crest development leads to the branchial arch syndromes.
- Numerous signaling systems are involved in cranial suture morphogenesis and fusion.
- Defects in the cranial base underlie the facial dysostoses.
- Genotype-phenotype correlation in the FGFR-related facial dysostoses is complicated by their promiscuous signaling mechanism.
- Management of any craniofacial patient can be performed systematically by breaking issues down into airway, feeding, vision, hearing, and speech.

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19

Unilateral Cleft Lip Repair

William Y. Hoffman • David M. Fisher



left lip repairs have been documented for more than 3000 years, but only in the past half century has cleft care become more team oriented, with better and more predictable outcomes. To some extent, these surgical advances have corresponded to advances in pediatric and neonatal intensive care and anesthesia, but beyond, a gradual and incremental addition of technical improvements has resulted in much-improved appearance and function in patients with cleft lips.

The incidence of cleft lip and palate varies among different populations, from approximately 1 in 300 to 1 in 1000; approximately 50% have a cleft lip and palate, 30% cleft palate only, and 20% cleft lip only. Cleft lip is more common on the left side; the ratio of left/right/bilateral clefts is 6:3:1. Right-sided clefts are more often associated with syndromes.¹ A genetic or racial predilection for clefting has been noted; the incidence appears to be highest in Asian and Hispanic populations. A native tribe in Venezuela has an incidence of approximately 25%.² Numerous genetic syndromes are associated with clefts.

Although right-sided complete cleft lip may be more commonly associated with a syndrome than left-sided or bilateral cleft lip, cleft palate alone is still seen more commonly in association with syndromes than cleft lip plus or minus cleft palate. Regardless, children with cleft lip and palate should undergo genetic evaluation, because this may reveal more subtle associated anomalies and be predictive of possible recurrence rates. *Van der Woude syndrome* is an autosomal dominant condition in which clefts (with variable expression) are associated with lower lip “pits,” which are sinus tracts of the minor salivary glands.³

Cleft lip can be diagnosed prenatally by ultrasonography as early as week 20 of gestation. Newer, three-dimensional ultrasound imaging techniques have been developed, although not yet in widespread use, that provide a virtual image of the face; a number of studies have demonstrated the remarkable abilities of these studies to show congenital facial abnormalities^{4,5} (Fig. 19-1). In the future, if fetal surgery becomes more commonplace, it may be possible to repair cleft lips in utero with the advantage of the “scarless” healing that has been noted in the fetus.



Fig. 19-1 Three-dimensional ultrasound image of a fetus with a right cleft lip.

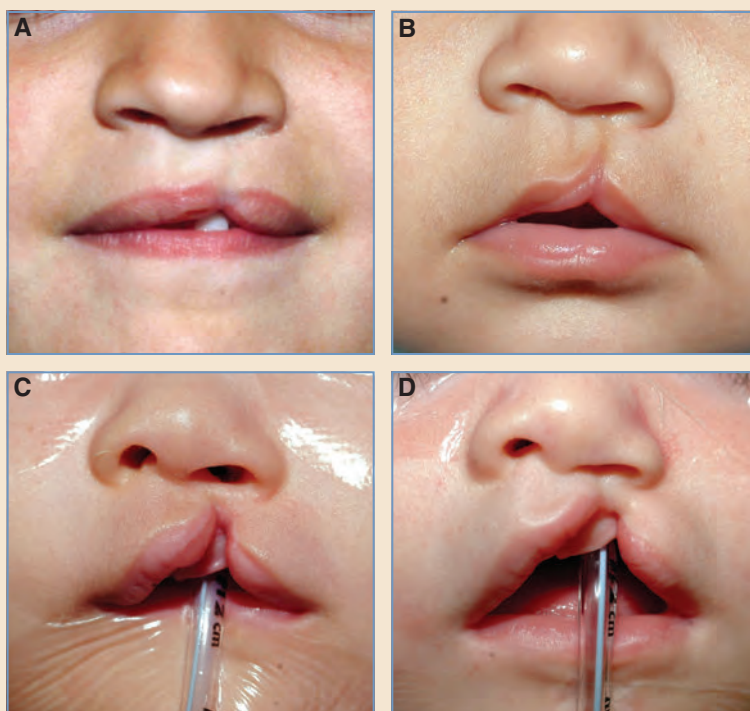


Fig. 19-2 Progression from a microform cleft lip to progressively greater incomplete clefts. **A** and **B**, Intact alveolar processes without a cleft palate. **C** and **D**, Greater involvement of the alveolus, with a complete cleft through the alveolus and palate shown in **D**. Greater nasal deformity is seen in **C** and **D**.

ANATOMY

Cleft lip results from a failure of merger of embryonic processes—the frontonasal process, which gives rise to the nose and the prolabium, and the maxillary processes on each side, which give rise to the rest of the upper lip on each side. The migration of neural crest cells from dorsal to ventral provides mesenchymal tissue, which is thought to supply the tissue to bridge the defects between the processes. The variable presentation of cleft lip is evidence that these events may be disrupted at various points in fetal development⁶⁻⁸ (Fig. 19-2).

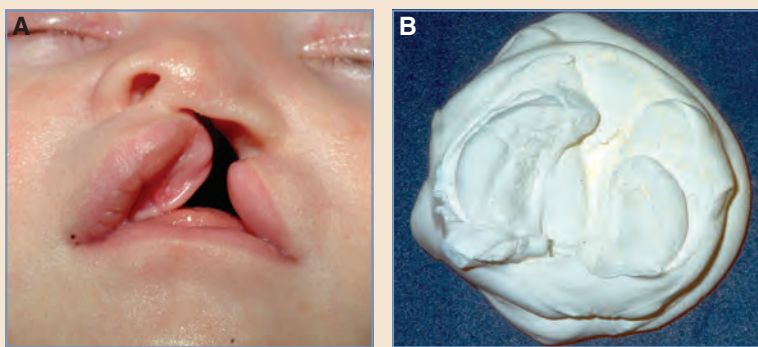


Fig. 19-3 A, Displacement of the alveolus caused by the pull of the orbicularis muscles contributes to the nasal deformity and to tension on the cleft repair. B, Dental models at the time of cleft lip repair show the displaced alveolar processes.



Fig. 19-4 A typical cleft nasal deformity in a patient with a microform cleft lip.

Some surgeons have postulated that in a cleft lip all of the normal structures are present but merely displaced, whereas others maintain that there is an absolute deficiency of tissue. The variability is wide, and probably each is true to a different extent in each patient. Incomplete clefts will not necessarily have better outcomes; an incomplete cleft with a paucity of normal tissue may be more difficult to repair than a wide cleft with an adequate amount.

An alveolar cleft contributes significantly to a cleft lip and nasal deformity. The abnormal insertion of the orbicularis muscle exerts a distracting pull on the alveolar segments, particularly the medial portion, creating a step-off between the segments and inherent asymmetry of the nasal base and tension on the lip repair (Fig. 19-3). If this is not addressed preoperatively, the lip repair will require stretching tissue over the protrusive premaxillary segment, making it more difficult to obtain adequate length of the lip on the repaired side and creating additional tension on the lip repair. The elevators of the upper lip are generally normal but must be respected during surgery; wide undermining may disrupt their normal attachments and produce an asymmetrical smile.

The degree of cleft nasal deformity varies. Several factors contribute to nasal deformity—even in an incomplete cleft, some maxillary deficiency may be present in the area of the piriform aperture on the cleft side. The septum is typically deviated, with a convexity obstructing the cleft-side airway and the caudal septum presenting in the noncleft side. The ala stretch, and the dome of the lower lateral cartilage slumps. After a cleft lip is repaired, nasal obstruction may occur on the cleft side because of the septal deviation (Fig. 19-4).

PREOPERATIVE ASSESSMENT AND NONOPERATIVE TREATMENT

An assessment of the overall health of the child is paramount. This begins at the first visit, preferably in the first week of life. Pediatricians may not see many cleft patients in their practices. Particularly for children who have a cleft palate together with a cleft lip, it is imperative that a feeding specialist see the family to ensure that the infant is receiving adequate nutrition. A palatal cleft will reduce an infant's ability to suck, and most patients are not able to breast-feed.

Special bottles and nipples facilitate feeding infants with a cleft palate, and evaluation by a nurse or occupational therapist with expertise in this area is essential. These children may also have swallowing problems and reflux that are not necessarily associated with clefting.

As previously noted, a genetic evaluation should be performed before lip repair, with a particular emphasis on other affected organ systems. Developmental delay is not generally a reason to delay a lip repair, unless a patient has significant hypotonia and recovery from anesthesia is a concern. Respiratory and cardiac problems would necessitate some delay in lip repair.

Most children with complete clefts benefit from some sort of manipulation of the alveolar segments before cleft lip repair. A number of centers use molding plates to move the segments into a more anatomic relationship; the addition of a prong to mold and stretch the nasal cartilage is called *nasoalveolar molding*.^{9,10} This has the advantages of reducing tension on the lip repair and greatly reducing the cleft nasal deformity (Fig. 19-5). For patients whose alveolar segments are in close contact, gingivoperiosteoplasty has been proposed as a method to reduce the need for later bone grafting and to maintain the maxillary segments in position after the molding process. The disadvantages of this procedure are the labor involved and the need for frequent office visits, which may be a problem when patients have to travel a long distance to be seen by a cleft team. Long-term outcome studies are in preliminary stages but appear favorable; however, at least one study has suggested that the nasal improvement from nasoalveolar molding tends to regress with time after lip repair.¹¹

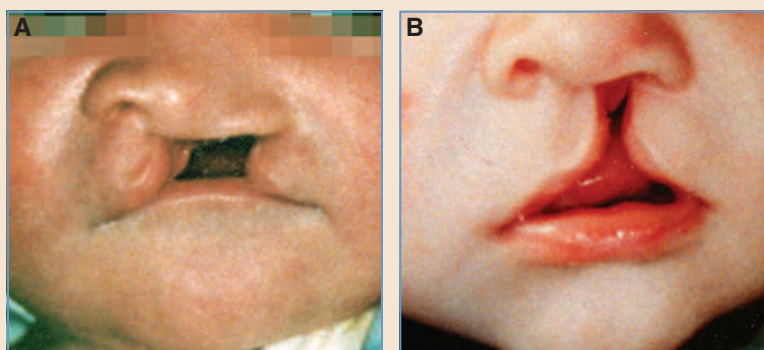


Fig. 19-5 A, A unilateral cleft lip before nasoalveolar molding. B, The same patient after nasoalveolar molding. (Courtesy of Barry Grayson, DDS, and Court Cutting, MD.)



Fig. 19-6 Presurgical taping is a low-cost method of closing an alveolar gap, although with less accuracy than nasoalveolar molding.

Other alternatives to nasoalveolar molding include external pressure, either with elastic devices or more simply with tape pulled across the cleft tightly enough to reduce the alveolar gap (Fig. 19-6). The advantages are simplicity and low cost, without the need for close follow-up; however, the gap does not close to the same extent that can be achieved with molding, and thus gingivoperiosteoplasty is not generally possible.¹² Whether this is an important factor will only be determined by prospective outcome studies of specific treatment protocols.

Lip adhesion is another technique that has been applied to narrow alveolar gaps. In contrast to nasoalveolar molding or taping, this is a surgical procedure that basically converts a complete cleft to an incomplete cleft by suturing relatively small subcutaneous flaps together.¹³ The disadvantages of this technique include scar tissue in the area of the definitive cleft lip repair and the risk of dehiscence of the small flaps, which is higher with wide clefts where the need for some sort of control of the alveolar gap is greatest.

SURGICAL OPTIONS

Many surgical methods for cleft lip repair have been described, and new modifications are reported on a regular basis. Early repairs simply pared the cleft margins and sutured them together¹⁴ (Fig. 19-7). A straight-line repair is used for only the most minimal of clefts; otherwise, scar contracture will shorten the lip and to some extent re-create the deformity. All other lip repairs have in common the use of local flaps to elongate the lip at the high point of Cupid's bow and to disrupt the forces of scar contraction. In the early part of the twentieth century, LeMesurier¹⁵ described the use of quadrilateral flaps for this purpose, but these techniques required discarding a substantial amount of lip tissue and disrupted the normal philtral anatomy (Fig. 19-8).

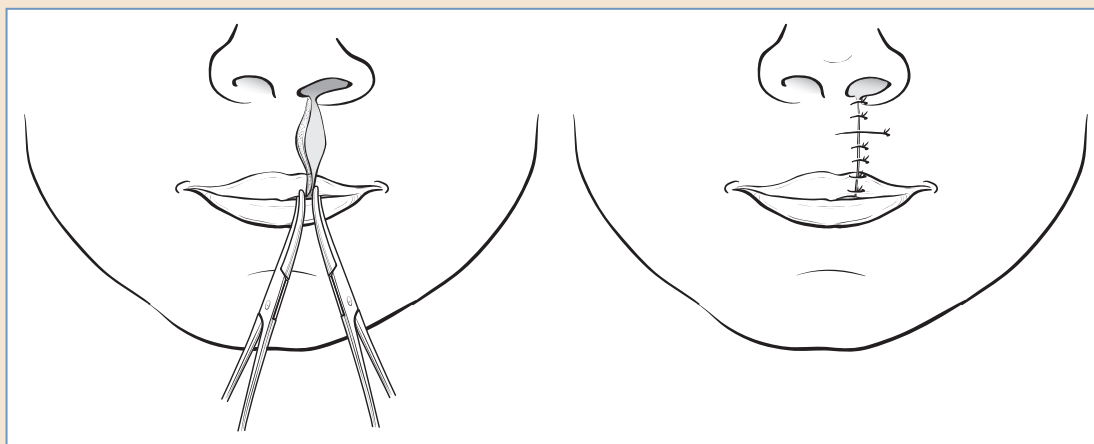


Fig. 19-7 Straight-line repair.

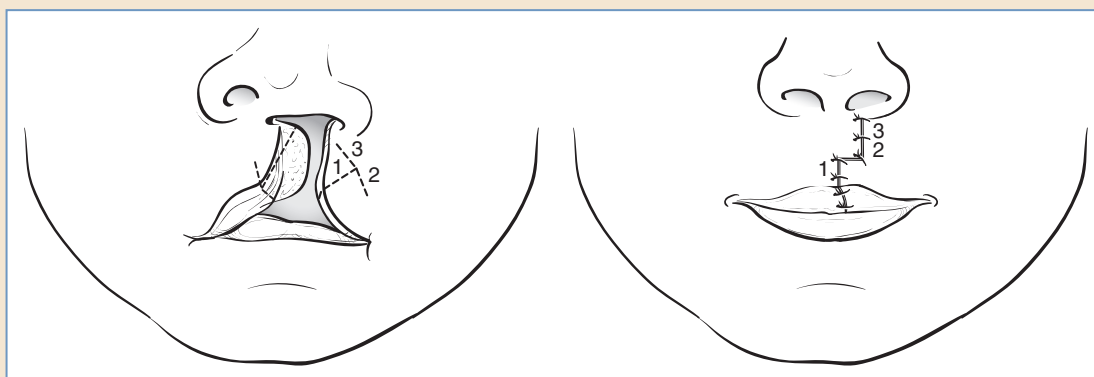


Fig. 19-8 LeMesurier repair.

A triangular lip repair (Tennison-Randall)^{16,17} changed the right-angle flaps of a quadrilateral repair to a modified form of Z-plasty just above the vermilion border (Fig. 19-9). An *inferior triangle repair* is a Z-plasty in which the middle limb of the A is shared by the incised cleft margins. Tennison¹⁶ used a bent wire as a stencil to produce a zigzag scar with equal limbs. Randall¹⁷ used calipers and simple mathematics. He created the required length while reducing the size of the inferior triangle. In this repair, the total lip height of the noncleft side is measured from a reference point in the nostril sill to the noncleft-side peak of Cupid's bow. The greater lip height is measured on the cleft side from the proposed medial point of closure at the height of the lip in the nostril sill (symmetrical with the reference point in the noncleft-side nostril sill) to the cleft-side peak of Cupid's bow. The difference between the total lip height and the greater lip height is the approximate height of the lesser height, the base width of the inferior triangle required to level Cupid's bow (see Fig. 19-13). An inferior triangle repair has several advantages. The nostril sill is closed by simple side-to-side approximation of medial and lateral nostril sill elements, and scarring at the base of the nose is minimal. Lip length can be obtained even when the medial lip

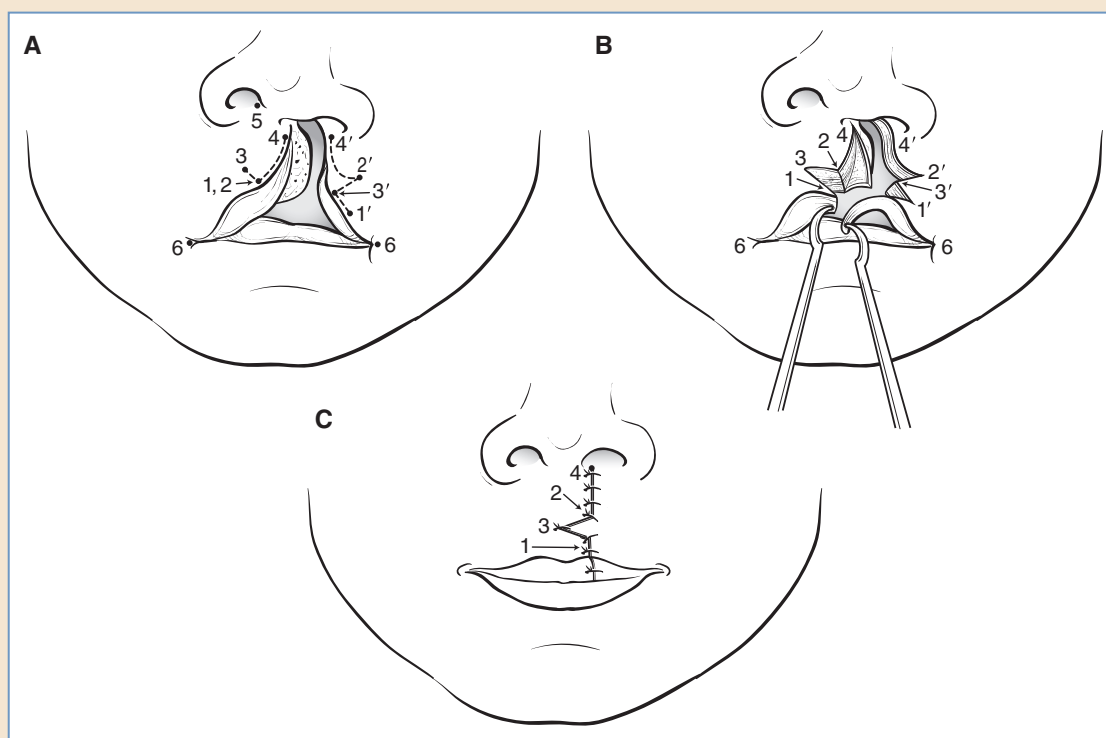


Fig. 19-9 Tennison repair. **A** and **B**, Markings for a triangular (Tennison) repair. Numbers 1 through 4 will be joined to their counterparts in the repair. **C**, Length is increased from the Z-plasty placed in the lower part of the philtrum.

height is very short. When the lateral lip height is short, often the case in patients with a complete cleft, lateral lip height can be added without compromising *Noordhoff's point* (identified on the vermilion-cutaneous junction, where the cutaneous roll and vermilion-mucosal junction lines start to converge medially).¹⁸ The disadvantage is a nonanatomic zigzag scar. Secondary revisions may be challenging, because the Z-plasty is in the middle of the lip.

In Millard's initial rotation-advancement repair,¹⁹ a curvilinear (rotation) incision is made in the medial lip element. The rotation incision mirrors the noncleft-side philtral column in its lower half. The incision then skirts along the lip-columellar crease and approaches but should not cross the noncleft-side philtral column. The incision allows caudal rotation of the cleft-side peak of Cupid's bow. The resultant defect is filled with a large, triangular (advancement) flap from the lateral lip element. Prolabial skin lateral to the rotation incision forms a C flap. It is rotated laterally and contributes to closure of the nostril sill (Fig. 19-10).

Millard²⁰ observed that the rotation of Cupid's bow was often inadequate and introduced a backcut extending from the most cranial extreme of the rotation incision down the lip just medial to the noncleft-side philtral column. The backcut is extended caudally as much as needed to level Cupid's bow. It leaves a quadrilateral-shaped defect that is filled by the C flap. The C flap can be advanced to elongate the columella on the cleft side.

The main advantage of a rotation-advancement over previously described repairs is that it preserves Cupid's bow and the philtral dimple. At least in the lower half of the lip, the scar mir-

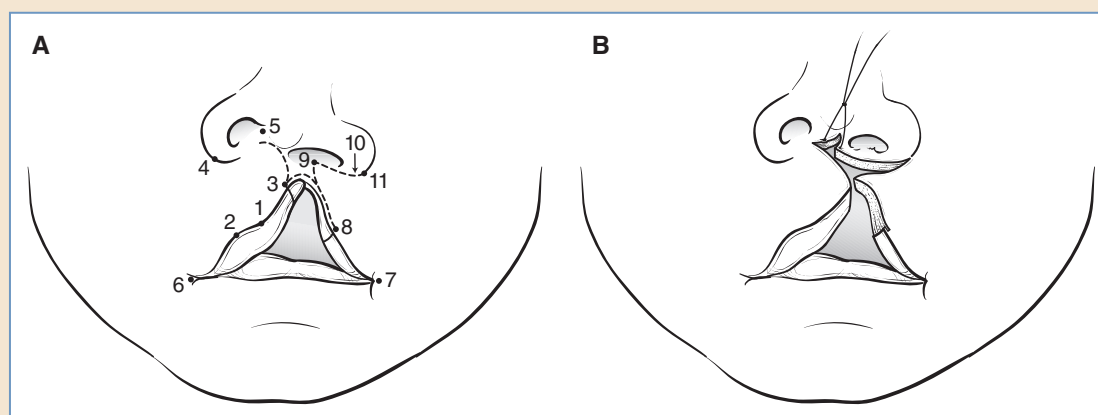


Fig. 19-10 Millard repair. Flaps are cut and ready for transposition; the Z-plasty is moved under the nasal sill. **A**, Advancement flap incisions are marked along the edge and around the alar base. A C flap is present under the columella. **B**, Advancement of the C flap into the columella. In Millard's original description, lateral placement of the C flap created a Z-plasty of the vertical scar that placed the tension just under the nasal sill and columella, creating flatness where it is normally seen and allowing a more natural pout of the lip at the vermilion border.

rors the noncleft-side philtral column. Many surgeons like the so-called cut-as-you-go execution of the repair.

Rotation-advancement has several drawbacks. The scar is not the anatomic mirror image of the philtral column in the upper half of the lip. The repair introduces excessive scar at the base of the nose (columellar base, nostril sill, and alar crease). When the C flap is used to fill the backcut and support the columella, it no longer can be used to the same extent to close the nostril sill. Nostril stenosis can result. The medial and lateral elements of the nostril sill are present in the unrepaired state. Augmenting the nostril sill closure with skin from the lip is not needed, and the C flap only interrupts the nostril sill roll. A long incision around the base of the nose is unnecessary and produces an obvious scar. Many surgeons have abandoned this element of the repair, deeming the alar crease incision unnecessary. The length of the cleft marginal incision of the lateral lip must be sufficiently long to meet with the full length of the rotation incision. When the lateral lip element is vertically short (as in most complete clefts), surgeons will need to extend the incision beyond Noordhoff's point. The transverse length of the lateral lip, usually already short, is thus further compromised to obtain vertical height.

Mohler²¹ altered the markings of the rotation-advancement repair to result in a scar that is more symmetrical with the noncleft-side philtral column. The change was made by straightening the curve of the rotation incision and extending it into the columella. The backcut was turned 90 degrees and ended at the lip-columellar crease. The resultant transverse limb of the repair is elevated to the lip-nose junction, marking an improvement over the original version. Mohler stated that the columella seems to be a rather silent donor site; however, Noordhoff¹⁸ warned that when the columella is narrow, a Mohler repair is contraindicated. The advantage of the repair over Millard's rotation-advancement is a more anatomically positioned scar. However, it shares with all rotation-advancement repairs a necessary compromise of transverse lateral lip length when the lateral lip is vertically short.

Noordhoff¹⁸ modified Millard's original repair. In this modification, an alar crease incision is not used. When rotation of Cupid's bow is inadequate, a small opening cut is made above the cutaneous roll above the cleft-side peak of Cupid's bow. A small triangular flap from the lateral lip is introduced into this defect. This triangle is inconspicuous and provides tension in the repair in the ideal position, above the roll, which accentuates the pout of the lip. Noordhoff also stressed the importance of achieving balance in the red lip and a level vermilion-mucosal junction (red line). Vermilion is almost always deficient in height below the cleft-side half of Cupid's bow. Here, Noordhoff augments the deficiency with a laterally based vermilion flap from the lateral lip.

I (D.M.F.) described the *anatomic subunit approximation technique*,²² which aims to produce a cutaneous scar along the seams of anatomic subunits. It is a modification of the Rose-Thompson^{23,24} repairs and usually incorporates elements of a modified inferior triangle repair. It offers several advantages over previously described techniques. (See Surgical Technique: Fisher Anatomic Subunit Approximation for a more detailed discussion.)

Management of the alveolar cleft was mentioned previously. If the alveolar segments can be brought into proximity, a gingivoperiosteal flap can be elevated and closed in continuity with the lip repair. The effects on subsequent maxillary growth and dental eruption patterns are concerns with this procedure. Follow-up of small cohorts of patients has shown that bony union of the alveolus occurs in most patients, and that approximately two thirds of the patients do not require subsequent bone grafting.²⁵ Growth appears to be similar to that in patients with an unrepaired alveolus. A small number of centers place a rib graft in the alveolar gap in the first year of life and claim that their patients have no growth retardation.²⁶ A comparative study showed some deficiency in maxillary growth in patients undergoing early bone graft, compared with gingivoperiosteoplasty or no early alveolar closure.²⁷

The level of undermining of the medial and lateral flaps during repair has been a topic of some discussion. The traditional method of repair has used undermining in the soft tissue to release the orbicularis muscle, leaving the periosteum intact. Delaire et al²⁸ and more recently Carstens²⁹ have advocated subperiosteal undermining with the argument that it restores normal anatomy more naturally and follows embryonic field anatomy. Although this is theoretically appealing, there are no long-term data or comparative studies on this issue. Older animal studies suggested that subperiosteal dissection may result in greater restriction of maxillary growth. Whether active or passive appliances are used to aid in approximation of the alveolar segments before lip repair, the need for extensive undermining of any kind is minimized, and the risk of growth restriction is minimized to the extent possible.

As lip repair has become more predictable, the nasal deformity is recognized as a persistent stigmata of cleft patients. McComb and Coghlan³⁰ first popularized correction of the cleft nasal deformity at the time of the initial lip repair by demonstrating long-term outcomes without deleterious effects on nasal growth. Preoperative nasoalveolar molding may possibly supplant surgical nasal correction, but some patients will benefit from some correction of the nose at the time of lip repair.^{25,31}

SURGICAL TECHNIQUE: HOFFMAN

Dedicated pediatric anesthesiologists are critical. Anesthesia is induced by mask and intravenous access is established. No antibiotics are needed for a soft tissue operation. An oral right-angle endotracheal tube is ideal for this procedure and is secured in the midline of the lower lip and chin. An oral gauze pack maintains the tube in a stable position, reduces leakage of gas from the uncuffed tube, and prevents blood from entering the posterior airway. The eyes are protected with clear plastic adhesive dressings, and the entire face is prepared with povidone-iodine solution.

The lip is marked with brilliant green dye, which is alcohol based and is useful for making fine lines; a fine skin pen can be used as well. The marking must be made before infiltration of a local anesthetic. Once the local anesthetic is injected, the lip is distorted, making evaluation of the anatomy more difficult. Visualizing a normal lip and constantly referring to the opposite side to guide the repair help to ensure symmetry, which is essential for a good outcome.

A rotation flap is marked first. The new high point of Cupid's bow on the cleft side is determined by first marking the midline and then the existing high point on the normal side; this distance is matched on the cleft side. This point on the vermilion border is tattooed using a 27-gauge needle and brilliant green dye. Surgeons tend to make this point too far up along the cleft margin, which is more lateral. This will result in greater difficulty in establishing adequate rotation downward and will produce a prolabium that is too wide. The rotation flap curves from the tattoo mark on the vermilion border upward to the base of the columella. The upper portion of the rotation flap should not cross the opposite philtral column; if necessary, a backcut along this line can be made for additional rotation. The remaining cutaneous portion of the medial lip, between the rotation flap and the mucosa, becomes the C flap. The mucosa is cut directly down from the vermilion border at the high point of Cupid's bow, leaving a base at the premaxilla. No mucosa should be included in the C flap, because this will cause the scar to have a permanent pink color.

The advancement flap results from the separation of the mucosa and the skin on the lateral lip segment. Many methods are available for determining the point along the vermilion border where the lateral and medial lip elements should meet. The greatest fullness of the mucosa has been advocated, as well as a point where the white roll becomes well defined. In general, because the height of the advancement flap from the vermilion border to the alar base will determine the lip height, I (W.Y.H.) prefer to measure the corresponding height on the contralateral side and transpose this measurement to the cleft side. This is another example of the principle of using the normal side as a reference. As on the medial side, the vermilion border is tattooed to mark this spot once it has been determined. As mentioned previously, the mucosa should not be included in the cutaneous flap; doing so permanently discolors the scar (Fig. 19-11).

The lip is infiltrated with lidocaine 0.5% and epinephrine 1:200,000. This mixture has 5 mg of lidocaine and 5 µg of epinephrine per milliliter of solution, and if no more than 1 ml/kg is used, the dosage will be well within acceptable limits. Ideally, a full 7 minutes is allowed for the hemostatic effect of the epinephrine, which will be apparent from the blanching of the soft tissue.

A rotation flap is incised first, all the way through the vermilion border down to the premaxilla. A C flap is cut, and a medial mucosal flap is elevated with the submucosal glands down to the premaxilla to expose the underlying orbicularis oris muscle. The C flap is dissected upward superficial to the orbicularis muscle, which is just below the dermis. The dissection of the orbicularis muscle is circumferential but does not extend farther than the midline of the prolabium to preserve the natural philtral dimple. The plane of dissection is just below the dermis in the cutaneous portion, but is somewhat deeper in the mucosal portion, preserving the submucosal glands with the mucosal flaps. The muscle is released from the base of the columella. This allows downward rotation of the muscle and the cutaneous flap. Last, the muscle is freed from the premaxilla itself, staying above the periosteum.

The advancement flap is incised in a similar manner; again, the mucosal flap is rotated downward and based on the maxillary attachment, and the orbicularis muscle is dissected circumferentially. Unlike Millard's initial description,¹⁹ no incision is made around the alar base; this is unnecessary and creates a difficult scar with permanent effacement of the alar-cheek junction. A small tongue of dry mucosa is maintained on the advancement flap, because usually the central prolabium contains an insufficient amount of dry mucosa. Once the mucosal flap is dissected down to the periosteum, the advancement flap is released along the vestibule, extending this incision

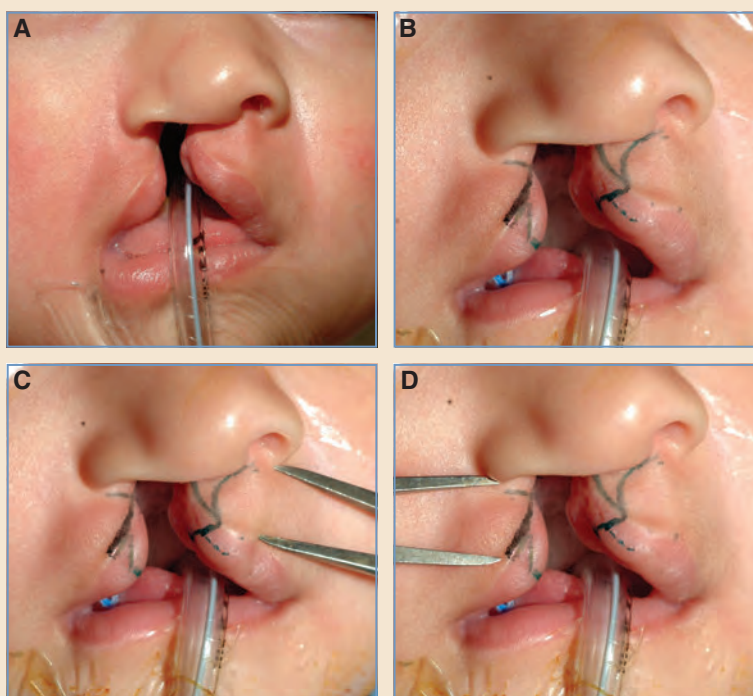


Fig. 19-11 A, A complete cleft lip before markings are made. A right-angle endotracheal tube is secured to the lower lip. B, Markings are made with brilliant green dye. C, The height of the lip is measured on the uninvolved side. D, The height is matched to determine the length of the advancement flap on the lateral lip element.

approximately 1.5 cm and then dissecting above the periosteum on the face of the maxilla. This is done entirely with blunt dissection; a finger on the inferior orbital rim will protect the eye.

A McComb nasal correction is started before closure.³⁰ First, the nose is released from the piriform aperture. The dissection on the face of the maxilla allows the scissors to be placed on the piriform aperture with one edge on the inside of the nose. The lateral nasal wall is sharply divided just above the inferior turbinate. A small tenotomy scissors is used to dissect over the alar cartilage on the cleft side of the nose, entering below the C flap in the midline and from the alar base laterally. This dissection is carried over the domes of both alar cartilages and up onto the dorsum. Releasing the lateral crus of the alar cartilage on the cleft side all the way down to the alar rim is critical (Fig. 19-11, E through G).

The lip closure progresses from inside to outside. The lateral mucosal flap is transposed 90 degrees to close the defect on the nasal sidewall. Usually, some of the length of this flap must be trimmed to fit well. The medial mucosal flap is turned over and sutured to the medial portion of the lateral flap. This provides lining for the floor of the nose and the vestibule of the advancement flap as it is brought across the cleft (Fig. 19-11, H). The nasal sidewall is now closed to the newly created floor of the nose, suturing to a small portion of the lateral mucosal flap and then to the medial mucosal flap, with the corner of the nasal cut coming into the defect left by the top of the C flap. Comparison with the opposite side of the nose provides a template to establish

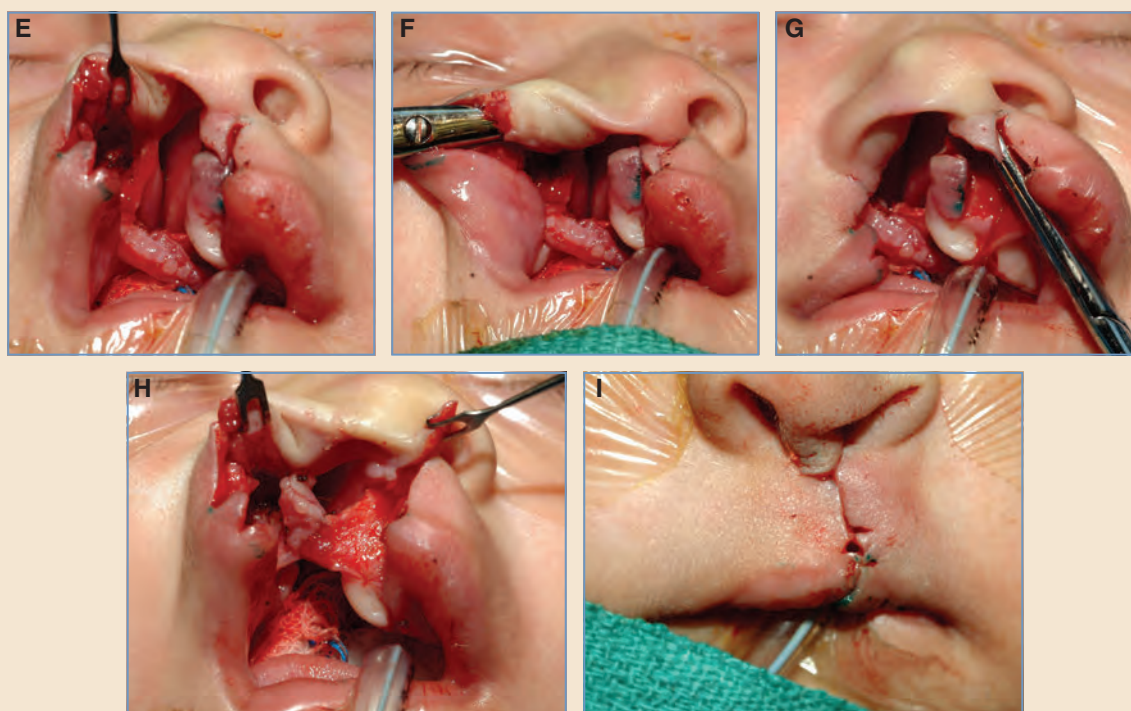


Fig. 19-11, cont'd E, The nose has been freed from the piriform aperture, together with the advancement flap. F, Freeing the alar cartilage from the overlying skin from the lateral incision. G, The alar cartilage is further released from the midline. H, The nasal floor is closed with the lateral and medial mucosal flaps sutured to each other across the alveolar cleft. I, A triangular lip flap is performed just above the vermilion border to lengthen the repair.

symmetry. The vestibular incision is closed, placing medial traction on the advancement flap to determine placement of the sutures.

The alar base is anchored to the base of the columella using an absorbable monofilament suture. This has the dual function of taking tension off the nasal floor repair and bringing the tilted columella into the midline. The orbicularis muscle is also closed with absorbable monofilament suture; this is a critical repair, because the integrity of the muscle is linked to subsequent lip growth and to normal function. Rotating the medial muscle downward is essential; these sutures should take tension off the lip repair, and the initial tattoos on the vermilion border should be aligned at this point.

The lip closure begins by rotating the C flap medially between the base of the columella and the rotation flap. This takes some of the tension off the advancement flap, adds to the height of the repaired side of the lip, and creates better symmetry of the columella. If the rotation and advancement flaps overlap, minimal trimming can be done; the curve of the final closure should be a mirror image of the opposite philtral column as much as possible. Skin is closed with 6-0 fast-absorbing catgut; occasionally, two or three dermal sutures of 5-0 material are placed to relieve tension, but usually this is achieved by the muscle repair. After the C flap is closed, the first suture is placed through the two tattoos at the white roll.

The length of the repaired lip must be carefully assessed. If the repair has adequate length, the mucosal closure should be completed; otherwise, a triangular lip flap, as described by Noordhoff, should be used. This involves a 1 to 1.5 mm incision just above the vermilion border

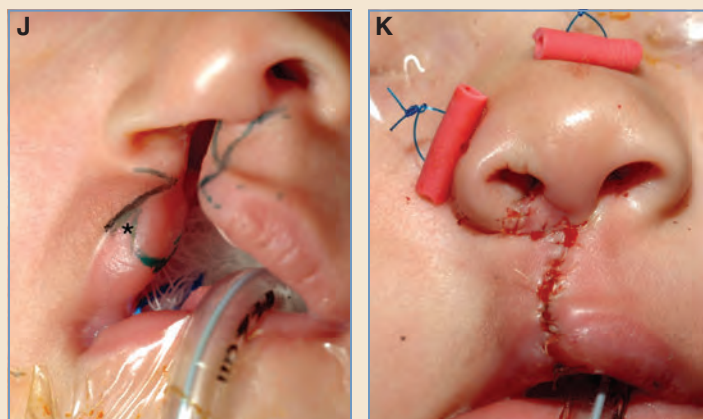


Fig. 19-11, cont'd **J**, A flap of dry vermilion (*) is preserved from the lateral lip element, which will be inserted into the medial lip to improve the balance of wet and dry mucosa. **K**, The completed lip repair with bolster sutures placed. The C flap is positioned in the nasal sill in this patient, although it is often rotated medially under the columella.

on the medial side and a corresponding small flap to fill the defect from the lateral side. Essentially, it is a tiny Z-plasty placed just above the vermilion border (Fig. 19-11, *I*). The only contraindication to using this flap is a lip that is so short that the surgeon thinks a revision will be required once the child has grown. In these cases, surgeons may choose not to use a triangular flap at the initial lip repair and to wait until the revision.

The mucosal repair is equally important. The alignment of the dry vermilion and wet mucosa, the so-called red line, is assessed first. As noted previously, a paucity of vermilion in the central part of the lip is typical. By preserving a tongue of vermilion with the lateral advancement flap, this can be inserted into an incision made along the vermilion medially (Fig. 19-11, *J*). Any excess below this is trimmed, and a 5 mm Z-plasty is placed in the wet mucosa to prevent notching of the closure.

The nasal correction is completed after the lip is closed. Two bolster sutures are placed in the nose to create a new relationship between the alar cartilage and the overlying skin. The first is placed in the dome of the alar cartilage and brought out percutaneously at the opposite side of the dorsum to pull the alar dome up and medially. The second is a percutaneous suture placed across the fold of the lateral crus that inevitably occurs, exiting in the lobular crease of the nose. Both of these sutures are polypropylene and are applied with pieces of 8 Fr rubber catheter to bolster the skin against erosion. The sutures are tied relatively loosely, and the tension of the sutures on the rubber is used to keep the repair in place. Generally, this appears overcorrected in the operating room and often appears undercorrected later (Fig. 19-11, *K*).

Postoperative Care

Bacitracin ointment is placed on the suture line. We prefer to place a Logan bow to relieve tension and to protect the lip repair for the first few days; this is not replaced once the tape loosens. Others think that this device is not needed to protect the lip repair. Arm restraints are used for approximately 10 days. Most of our patients stay in the hospital overnight, because they come from too great a distance to drive home, but local patients may be discharged the same day if they are doing well. No feeding restrictions are necessary, and whatever method of feeding was used preoperatively is continued after surgery.

Patients are seen in the office 4 to 5 days postoperatively. The fast-absorbing catgut sutures are absorbed in this short time, thus preventing crosshatch marks and the need for suture removal. The two nasal sutures and bolsters are removed.

Outcomes and Complications

Complications of cleft lip surgery are rare. Although opinions vary between surgeons, neither one of us gives patients prophylactic antibiotics for cleft lip repairs. Out of nearly 1000 cleft lip repairs, none has had an infection, and only one had a dehiscence. A small number of patients developed inclusion cysts, and no hematomas occurred.

Typically, scar hypertrophy begins after 3 to 4 weeks, and parents are informed about this in advance. Although taping or massage may improve scars, it can be difficult to perform in an infant. Generally, scar hypertrophy improves on its own. If the lip had adequate length at the end of the procedure, it may appear a little short as the scar hypertrophies, but it usually returns to a symmetrical length after several months (Fig. 19-12).

The goal of a unilateral cleft lip repair should be a normal appearance with symmetry of the lip and nose. Minimal or no revision of the lip should be required. Usually, something can be found to be lacking, but it should be possible to delay revision until bone grafting is performed or ideally until the patient is a teenager. The most common problem is redundancy of the mucosa on the cleft side, which results from movement of the alveolar shelves after the lip repair.

If a significant deformity persists after healing is completed, revision should be undertaken before the child is of school age for his or her psychological benefit. Such an early revision is required in less than 25% of our patients.

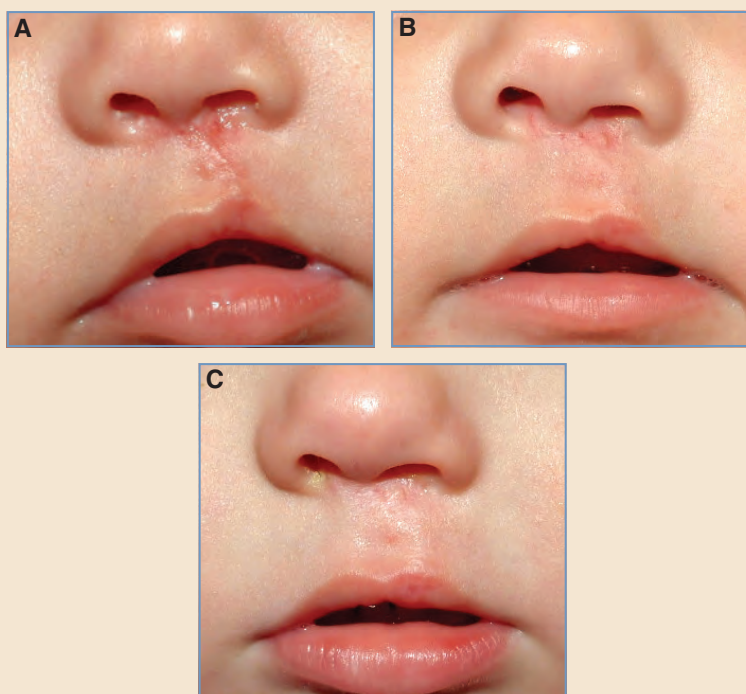


Fig. 19-12 A, Hypertrophic scarring 2 months after a cleft lip repair. B, Improvement in the scar and lip length 5 months postoperatively. C, Marked improvement 1 year postoperatively.

A nasal deformity is a different issue. The septum is deviated to the side of the cleft posterosuperiorly and toward the normal side caudally, and the dorsum is deviated away from the side of the cleft in most cases; the long-term results of nasoalveolar molding are not yet known in this regard. Even after a McComb procedure,³⁰ most if not all children with unilateral clefts require a septorhinoplasty in their teens; generally, earlier surgery on the septum may impede growth of the maxilla. Alternatively, the primary nasal correction described previously will markedly improve symmetry over no repair and will be sufficient for most children until they are teenagers.

Secondary surgery on the lip and nose is best delayed until the underlying bony and dental structures are as symmetrical as possible. Generally, this is after the first and second phases of orthodontics are completed, with alignment of the alveolar segments, bone grafting, and alignment of the permanent teeth into ideal occlusion. LeFort I maxillary advancement, if needed, should also be done before final lip and nose surgery.

SURGICAL TECHNIQUE: FISHER ANATOMIC SUBUNIT APPROXIMATION

Incisions for the anatomic subunit approximation technique²² cross the cutaneous rolls of the lip elements in a perpendicular fashion (Fig. 19-13). The incision on the medial cleft side then ascends from a point above the cutaneous roll to mirror the noncleft philtral column. An angle is formed between the incision line crossing the medial lip element roll and the philtral column

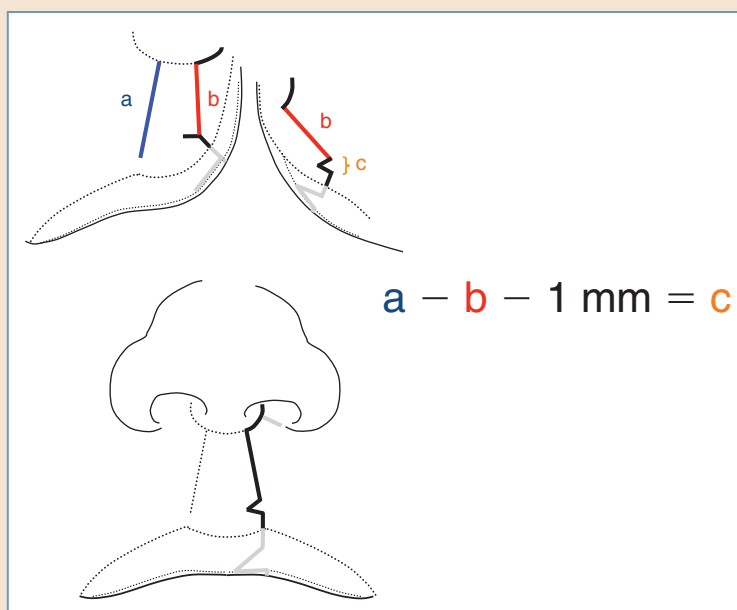


Fig. 19-13 Presurgical markings and measurements. The total lip height (*a*) and the greater lip height (*b*) are each measured from points above the roll to the base of the columella. The lesser lip height (*c*) (base width of the small, inferior triangle) is equal to the total lip height minus the greater lip height minus 1 mm. The 1 mm accounts for the Rose-Thompson lengthening that occurs as the angled incision lines approximate in the vertical.

$$\begin{aligned} \text{Lesser lip height} &= \text{Total lip height} - \text{Greater lip height} - 1 \text{ mm} \\ \text{Lesser lip} &= \text{Base width of the small inferior triangle} \end{aligned}$$

incision. This angle will open to create a straight line and lengthen the lip by approximately 1 mm. At the height of the lip, the incision line curves superolaterally, just below the lip-columellar crease and thus preserving it, to the medial point of closure in the nostril sill. Lip length is gained by two mechanisms: first, a Rose-Thompson^{23,24} effect as angled lines straighten on approximation (which accounts for approximately 1 mm of lengthening) and second, if required, a small inferior triangle from the lateral lip element, which is positioned just superior to the cutaneous roll. The lengthening obtained by following the Rose-Thompson principle significantly reduces the size of the inferior triangle that would otherwise be required had a traditional inferior triangle repair been performed. The technique also uses a Noordhoff lateral vermilion triangle flap (C flap) to augment the deficient vermilion height of the medial lip element on the cleft side.

This technique has many advantages over the inferior triangle and rotation-advancement repairs. In contrast to the rotation-advancement techniques,^{20,21,32} scarring at the base of the nose is minimal. The cutaneous scar is positioned along the seams of anatomic subunits with the exception of, when required, a small triangle above the cutaneous roll. For several reasons, this is the preferred position of an inferior triangle^{20,21,32}:

- Tension is ideally positioned above the roll and accentuates the pout of the lip.
- Continuity of the cutaneous roll is achieved by side-to-side approximation of the roll elements, simplifying the light reflex off the roll.
- It allows the creation of a lengthening angle above the roll.
- The roll does not need to be augmented—doing so will create a stairstep of the non-hair-bearing roll and hair-bearing skin junction. The repair preserves the lateral lip element on the cleft side.
- In common with the inferior triangle repair, lateral lip transverse length need not be compromised to gain vertical height, a commonly practiced compromise when performing a rotation-advancement-type repair.

The lateral lip element is almost always short in its transverse dimension, compared with the noncleft side. As commonly seen in complete clefts, the vertical height of the lateral lip is also short.^{33,34} The practice of moving the base of the incision laterally to gain vertical height is a compromise that is often required when performing a rotation-advancement repair, but is not required with the anatomic subunit approximation technique.

The cleft-side Cupid's bow peak and philtral column height are marked equidistant from the midline relative to the corresponding noncleft-side markings. The noncleft-side Cupid's bow peak is not marked at the highest point of the curve of the lip, but rather where the straight portion of the noncleft-side half bow meets the convexity of the lateral lip element (Fig. 19-14). No such convexity exists on the cleft side of the medial lip element. The corresponding convexity on the cleft side will come from the lateral lip element lateral to Noordhoff's point.

Points are marked above each bow peak just over the cutaneous roll at points above the lines perpendicular to the vermilion-cutaneous junction. The medial lip cleft-side philtral column incision (equivalent to the greater lip height) will ascend from the point above the roll to the height of the lip just below the lip-columellar crease. The incision line then traverses from the height of the philtral column incision (greater height incision) superolaterally to the medial point of closure in the nostril sill (this length is called the *third height*). From the point above the roll, the incision line continues caudally perpendicular to the roll through the bow peak and vermilion and across the free border of the lip into the mucosa.

The point of origin of the opening incision for the medial lip backcut (if indicated) is initiated just above the cutaneous roll. The opening incision is drawn perpendicular to the greater height incision.

The medial and lateral points of closure within the nostril sill are not distinct anatomic points and thus are the hardest points to define in this repair. The medial point of closure in the nostril

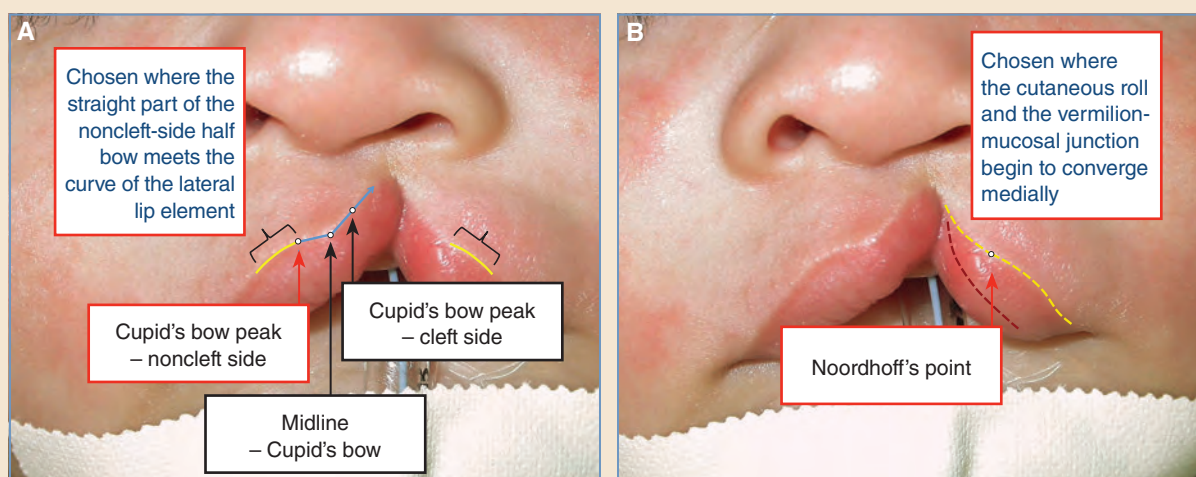


Fig. 19-14 **A**, The noncleft-side Cupid's bow peak is not marked at the highest point of the curve of the lip. It is marked where the straight portion of the noncleft-side half bow (*blue line*) meets the convexity of the lateral lip element (*yellow line*). There is no such convexity on the cleft side of the medial lip element. This curve of the bow on the cleft side will come from the lateral lip element lateral to Noordhoff's point, shown in **B**. The cleft-side peak of Cupid's bow is positioned on the vermillion-cutaneous junction on the cleft side of the medial lip element equidistant from the midline of the bow. **B**, Noordhoff's point is identified on the vermillion-cutaneous junction (*yellow dashed line*) where the cutaneous roll and the vermillion-mucosal junction lines start to converge medially. The upward convexity of the lateral lip element lateral to Noordhoff's point should be preserved.

sill is positioned lateral to the curve of the lip-columellar crease. It will be positioned more medially in complete cases and more laterally in incomplete cases. This will allow surgeons to adjust the length of the third height and therefore use less lateral lip height in complete cleft patients with vertical height deficiency or more of the lateral lip height in patients with incomplete clefts and excess vertical height. The lateral point of closure in the nostril sill is chosen on the lateral lip element, relative to the medial point of closure in the nostril sill. Approximation of these two points should accomplish two goals: the nares are of equal circumference and the alar bases are symmetrical from the anterior view. Gentle manipulation of the medial and lateral lip elements should confirm acceptable positioning of these points. In complete clefts in which the lateral lip is so tethered that the medial and lateral lip elements cannot be brought together, releasing the lateral lip element before marking the lip is advised.

Calipers are used to measure the *total lip height*, defined as the height of the noncleft-side philtral column from a point above the roll to a point just below the lip-columellar crease, and the *greater lip height*, defined as the height of the cleft-side philtral column incision from a point above the roll to a point just below the lip-columellar crease and positioned along a line that mirrors the noncleft-side philtral column (see Fig. 19-13). The total lip height is measured with the lip at rest. The greater lip height is measured with gentle downward traction on the lip to unfurl the medial lip. The lesser lip height (equivalent to the base width of the small inferior triangle) is equal to the total lip height minus the greater lip height minus 1 mm. The 1 mm accounts for the approximate length achieved by the Rose-Thompson principle. If an inferior triangle is required, the backcut for the opening incision is drawn perpendicular to the greater lip height incision and of a length equal to the lesser lip height (the inferior triangle is equilateral). Rotation of the bow is achieved by the combination of the backcut and the Rose-Thompson effect^{23,24} (as the angled lines straighten). The lesser lip height is usually 1 to 1.5 mm and never exceeds 2 mm.

For minor-form incomplete clefts, an inferior triangle is often not necessary. The backcut is made perpendicular to the greater height incision and not parallel to the cutaneous roll, which would produce a flattening of Cupid's bow when the backcut is opened. The perpendicular backcut preserves tissue above the roll and thus better conserves the curve of Cupid's bow.

The base of the cutaneous incision on the lateral lip element is marked according to the recommendations of Noordhoff. Noordhoff's point is marked along the vermilion-cutaneous junction at the point where the cutaneous roll and red line begin to converge medially. The natural upward convexity of the cutaneous roll lateral to Noordhoff's point is ideally suited for reconstruction of the cleft-side curve of the bow. A point is marked just above the cutaneous roll at a point above Noordhoff's point, along a line perpendicular to the vermilion-cutaneous junction. Between the lateral point of closure in the nostril sill and the point above the roll, three lengths must be accommodated: the greater lip height, the lesser lip height, and the third height (corresponding in length to the line connecting the height of the cleft-side philtral column and the medial point of closure in the nostril sill). This is readily achieved by placing the greater-lip-height caliper on the lateral lip element so that its lower tip lies the lesser-lip-height distance away from the point above the roll, and its upper tip lies the third-height distance from lateral point of closure in the nostril sill.

In complete clefts, with a short vertical height to the lateral lip element, the angle of the greater lip height incision on the lateral lip will be more horizontal and, in extreme cases, may be confluent with the upper limb incision of the inferior triangle (Fig. 19-15, *A*). It is also helpful in patients with a lateral lip element vertical height deficiency to position the medial point of nostril sill closure (and therefore the lateral point of nostril sill closure) more medially. This will shorten the third height, and as a result, less lateral lip element vertical height will be required to match this length. Conversely, in an incomplete cleft that presents a relative excess of vertical height, the third height can be lengthened by lateralizing the medial and lateral points of closure within the nostril (Fig. 19-15, *B*). More lateral lip vertical height can then be taken up by the longer third length. If vertical height excess persists despite this maneuver, it may be necessary to perform a medially based wedge excision of the upper lip below the lateral point of closure in the nostril sill (Fig. 19-15, *C*). The lateral apex of this wedge excision should never extend laterally beyond the subalare; this is unnecessary and leaves an unsightly scar. Care should be taken not to alter the vertical height of the lateral point of closure in the nostril sill, because it will compromise the final vertical position of the alar insertion.

A vermilion deficiency below the cleft-side half of Cupid's bow requires augmentation. An opening incision is created along the vermilion-mucosal junction below the cleft-side half of Cupid's bow. This backcut will receive a laterally based vermilion triangular flap from the lateral lip element, as described by Noordhoff.

In incomplete clefts, a wedge excision of the nostril sill above the medial and lateral points of nostril sill closure is performed. The excision should be sufficient to balance the circumference of the nares, with great care to prevent overresection, because the resultant nostril stenosis presents a difficult reconstructive challenge. The mucosa of the cleft margins is removed by wedge excision; a short, lateral, upper buccal sulcus advancement incision may be helpful.

In complete clefts, the mucosal incisions on the medial and lateral lip elements extend cranially to the points of attachment of each lip element to the greater and lesser alveolar segments, respectively. An upper buccal sulcus incision extending laterally from the point of attachment of the lateral lip element is made. A mucosal backcut can be incorporated as necessary. From the medial point of closure in the nostril sill, the incision continues intranasally along the caudal margin of the septum for a distance of up to 12 mm to facilitate access for septal repositioning. The lateral free edge of the septal flap will receive the medial free edge of the lateral vestibular

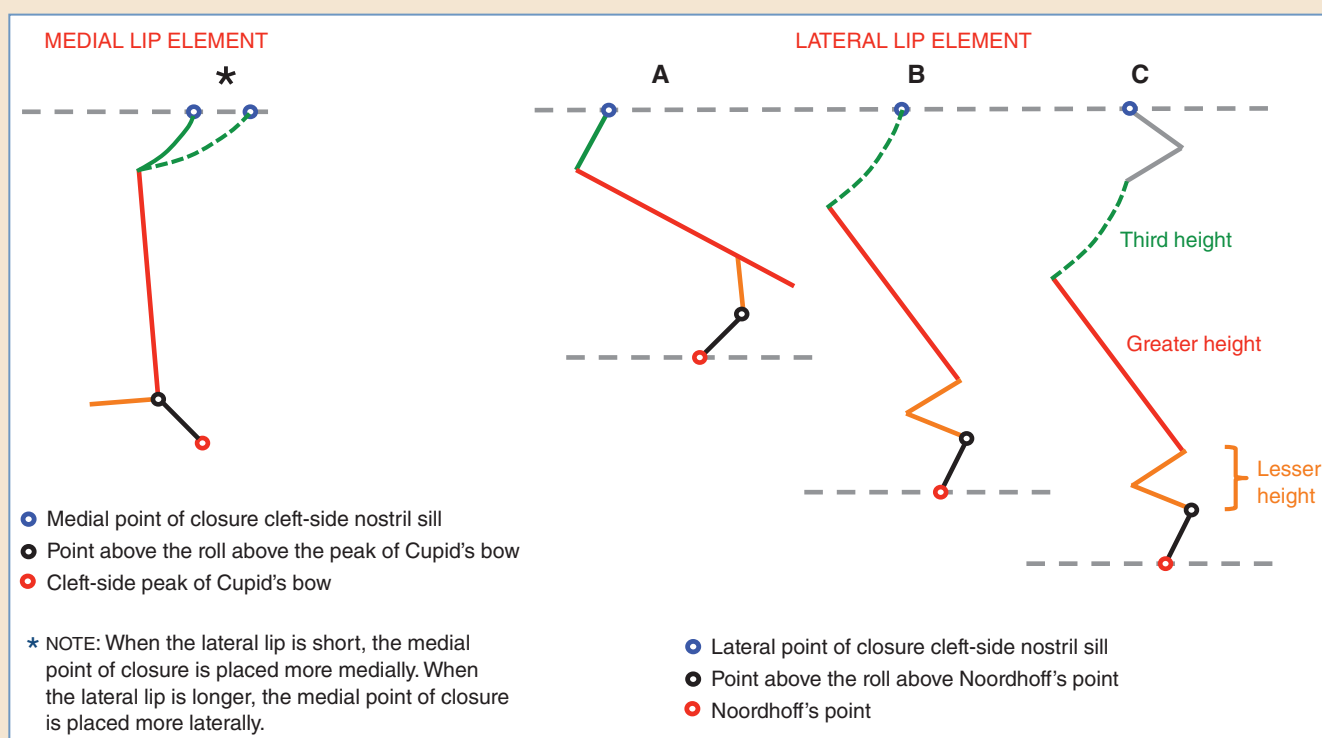


Fig. 19-15 Lateral lip markings will vary depending on the vertical height of the lateral lip element. **A**, In many complete clefts, the vertical height of the lateral lip element will be short. In such cases it is useful to position the medial and lateral points of nostril sill closure more medially, thus shortening the third height. The greater lip height incision on the lateral lip element will be more horizontal, and its lateral extreme may need to be coincident with the upper incision of the inferior triangle, which will require an upward-pointing orientation. **B**, With many incomplete clefts, the vertical height of the lateral lip element may be excessive. In this case, it is helpful to position the medial point of closure in the nostril sill more laterally. This essentially lengthens the third height. **C**, If the lateral lip remains too long despite this maneuver, a medially based wedge excision of the upper lip can be performed below the lateral point of closure in the nostril sill. The position of the point of closure in the nostril sill and of Noordhoff's point should not be compromised.

flap. The lateral vestibular flap is fashioned by incising from the lateral point of closure in the nostril sill to the attachment point of the lateral lip element to the alveolus and then along the piriform margin. This allows supraperiosteal mobilization of the alar base. An anteriorly based inferior turbinate flap may be incorporated to cover the vestibular lining defect in clefts with a significant AP distance between the greater and lesser maxillary segments.

Patients with a contralateral minor-form cleft should be considered for a synchronous bilateral cleft lip repair. This approach provides the best opportunity to achieve symmetry of Cupid's bow and of the resultant scar, and provides the ideal opportunity for reconstruction of the probial deficiencies of the cutaneous roll, vermillion, and median tubercle.

Primary Nasal Correction

The anatomic subunit approximation technique describes a technique for cleft lip repair. It can be accompanied by most previously described techniques of primary rhinoplasty.

Box 19-1 Goals for Primary Rhinoplasty

- Balanced alar bases (from the anterior view)
- Columellar base mobilization to the midline
- Caudal septal release and repositioning
- Alar base release and repositioning
- Release of the nasal attachments to the piriform margin
- Anteromedial advancement of the cleft-side dome and lateral crus
- Creation of the scroll—an internal nasal valve plication suture or sutures
- Lateralization of the cleft-side lateral vestibular lining—an alar transfixion suture or sutures
- No external skin incisions
- No excisions of the skin envelope

Because cleft nasal deformity is the result of an underlying skeletal deformity,^{35,36} the skeletal deformity needs to be corrected before a correction can be successful in the nose. Because pre-surgical orthodontics can only partially improve the deformity (by correcting maxillary segment malalignment while not addressing maxillary hypoplasia), and because primary bone grafting is not performed, completely correcting the nasal deformity at the primary operation is unrealistic. Patience is essential. The potential risks of a primary rhinoplasty include iatrogenic deformity of the skin envelope and cartilages and the introduction of scar that may complicate subsequent interventions. Skeletal base deficiencies can be augmented later at the alveolar bone grafting and orthognathic surgery if indicated. This will prepare the patient for definitive septorhinoplasty. Deformities such as a broad, deviated nasal dorsum, which are only obvious after maturity, may be addressed in this surgery. Definitive secondary septorhinoplasty will be successful only if the cartilages are intact and robust, and the skin envelope is complete and free of scar.

Our preferred primary rhinoplasty is relatively aggressive at the nasal base and relatively conservative in the lobule (Box 19-1).

A nasal stent is placed and secured with a transseptal suture. It is removed with the sutures after 1 week. The currently available stents do little to effect nasal tip position but are perceived to be helpful to smooth the nostril sill closure.

Management of the Cleft Alveolus

Our preferred method for managing the alveolar cleft involves presurgical nasoalveolar molding (for complete clefts), lip repair at 3 to 5 months of age (without gingivoperiosteoplasty), and secondary alveolar bone grafting before eruption of the adult cleft-side canine.

Surgical Technique

Local anesthesia is limited to bilateral infraorbital nerve blocks and for submucoperiosteal hydrodissection of the caudal septum. To prevent errors in measurement and in the perception of the balance of the repair, the lip elements are not infiltrated.

The repair should be performed in the proper sequence to allow the operator to assess progress (using the three checkpoints described in the following sections) and make modifications if necessary. The medial lip incisions (without the backcut) are made first.

First Checkpoint

The *first checkpoint* is the assessment of the length achieved by the Rose-Thompson effect alone. The 1 mm of calculated lengthening is an estimate. It will be greater when the slope of the bow is steeper: the more rotated the bow, the more acute the angle formed by the incision across the roll, and the greater the height incision, thus the greater the lengthening achieved as this angle opens to 180 degrees. Conversely, the more level the bow, the more obtuse the angle, and less the lengthening achieved by the Rose-Thompson effect. If the bow has been leveled by the Rose-Thompson effect, an opening incision and an inferior triangle will not be necessary. If an opening incision is deemed appropriate, it should be of sufficient length to level the bow.

Second Checkpoint

The *second checkpoint* is ensuring that the triangle that has been drawn on the lateral lip element is of the exact size to fill the defect created by the opening incision in the medial lip element once the bow has been leveled. The size of the triangle can be adjusted at this point if the lateral lip incisions have not already been performed.

Third Checkpoint

Once the medial and lateral lip elements have been fully mobilized from their skeletal attachments, the *third checkpoint* is performed—the approximation of the medial point of closure in the nostril sill with the lateral point of closure in the nostril sill. Before committing to the lateral point of closure, surgeons should check that the nares are of equal circumference, and that the cleft-side alar base has been sufficiently rotated to achieve symmetry with the noncleft-side alar base.

On the medial lip element, dissection between skin and muscle is limited to 1 mm from the incised margin to preserve the philtral dimple. Dissection between the skin and muscle on the lateral lip element is more extensive; skin is freed from the underlying muscle sufficient to advance the muscle relative to the skin and to release the bulge of the lateral lip. Muscle is approximated in a simple end-to-end fashion.

Absorbable monofilament sutures are used for the mucosa and muscular approximations. Skin is approximated with nonabsorbable sutures. Vermilion is approximated with braided absorbable sutures.

Postoperative Care

Elbow splints are worn for 2 weeks postoperatively. Patients may resume oral feeding immediately in the recovery room after surgery and are discharged on the following day if they are clinically well and oral intake is sufficient. Sutures are removed on the seventh postoperative day with the patient under general anaesthesia. The incision line is moistened with antibiotic ointment. Parents are instructed to keep the suture line clean and moist with Vaseline. A return visit is scheduled for 3 to 4 weeks postoperatively. Parents are given instructions regarding scar massage at this visit.

CASE EXAMPLES

This patient had a minor-form unilateral cleft and underwent an anatomic subunit approximation technique. In some cases, an inferior triangle is unnecessary, and the required lengthening can be obtained by the Rose-Thompson effect alone (Fig. 19-16).

This patient had an incomplete unilateral cleft and underwent an anatomic subunit approximation technique (Fig. 19-17).

This patient had a complete unilateral cleft lip and palate and underwent an anatomic subunit approximation technique (Fig. 19-18).

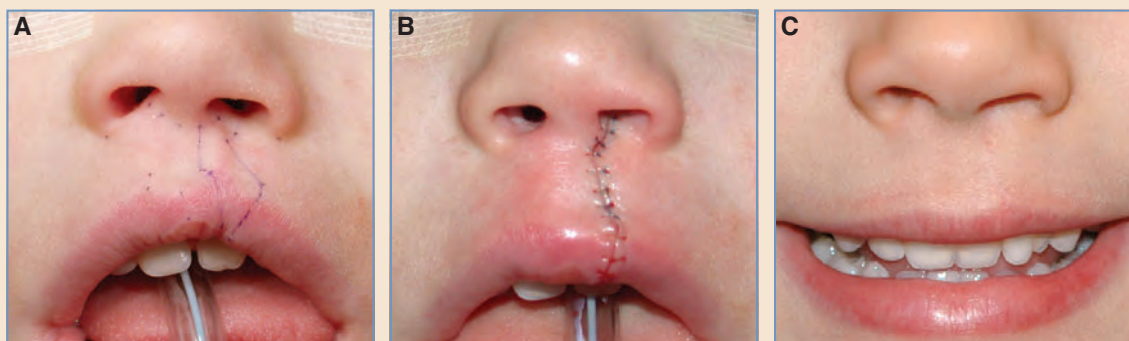


Fig. 19-16 A, The patient is shown preoperatively at 2 years of age. B, Immediate postoperative appearance. C, The patient is shown postoperatively at 5 years of age.



Fig. 19-17 A, The patient is shown preoperatively at 5 months of age. B, One-week postoperative appearance. C, The patient is shown postoperatively at 3 years of age.



Fig. 19-18 A, The patient is shown preoperatively at 6 months of age. B, One-week postoperative appearance. C, The patient is shown postoperatively at 5 years of age.

CONCLUSION

A unilateral cleft lip repair can provide reliable outcomes that minimize the stigmata of this congenital deformity. Many factors contribute to this achievement, most notably team care, which results in consistency in approach and careful follow-up. A balanced lip repair involves balancing the height deficiencies of the medial lip element with like tissue from the lateral lip element, while adjusting for the varying presentations of vertical height of the lateral lip element. Preserving the natural landmarks in both the medial and lateral lip elements will produce the most natural-appearing lip. Primary rhinoplasty should be performed judiciously and carefully to allow a definitive secondary septorhinoplasty. Although the techniques described in this chapter provide invaluable insight to understanding unilateral cleft lip deformities and have stood the test of time, experienced cleft surgeons may use other techniques with excellent outcomes. As with all reconstructive problems, evaluation and refinement of results in cleft patients are key elements for success.

KEY POINTS

Hoffman Surgical Technique

- Cleft lip can be diagnosed prenatally by ultrasound imaging as early as week 20 of gestation.
- Although syndromal associations are more common with isolated cleft palate than with cleft lip, a genetic evaluation is recommended for all children with cleft lip and palate.
- Reconstruction of the orbicularis muscle is critical to symmetry of the final lip repair and essential for normal lip growth.
- All lip repairs involve some degree of borrowing width to gain lip length, often in more than a single area of the lip.
- All key landmarks of the lip must be marked before infiltration of the lip with local anesthetic to prevent distortion of these landmarks.
- Mucosa should not be carried into the cutaneous flaps, because this will impart a permanent pink color to the scar.
- In a Millard type of cleft lip repair, the incision must not be carried around the alar base, because this will efface the alar-cheek junction. This is an extremely difficult problem to correct secondarily.

Continued

KEY POINTS (continued)

- In a primary nasal repair, releasing the lateral crus of the alar cartilage on the cleft side all the way down to the alar rim is critical.
- Nasoalveolar molding preoperatively may prevent the need for more extensive nasal dissection and release.
- Many authors advocate scar massage to decrease scar hypertrophy after cleft lip repair. How vigorously and frequently the parents perform this therapy will determine its effectiveness.
- In uncomplicated, isolated cleft lip repairs, attention to landmarks and all details of the repair will reduce the extent of secondary revision in most cases.
- Secondary surgery on the lip and nose is best delayed until the underlying bony and dental structures are as symmetrical as possible.
- Some degree of residual nasal asymmetry may be expected; generally, septorhinoplasty should be delayed until the time of skeletal maturity.

Fisher Anatomic Subunit Approximation

- The noncleft-side bow peak is marked on the vermilion-cutaneous junction where the straight part of the half bow meets the upper convexity of the lateral lip element. The cleft-side bow peak is placed an equal distance from the midline.
- A natural undulation of the cutaneous roll can only be achieved if the upward convexity of the lateral lip element lateral to Noordhoff's point is preserved.
- The transverse length of the cleft-side lateral lip element need not and should not be compromised to achieve vertical height.
- Leveling of Cupid's bow is the result of a Rose-Thompson lengthening effect in all cases. In most cases a small triangle is also placed above the roll.
- The orientation of the lateral lip element incisions vary depending on the vertical height of the lateral lip element.
- Vermilion deficiency below the cleft-side half of Cupid's bow should be augmented with vermilion from the lateral lip element.
- Medial incisions should be performed first. Lateral lip markings can be adjusted if necessary.

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20

Bilateral Cleft Lip Repair

Brad T. Morrow • Donald R. Mackay



he complete bilateral cleft lip (CL) poses multiple complex challenges to the reconstructive surgeon. The protruding premaxilla and the poor quality of the prolabial soft tissue make the goal of achieving a symmetrical tension-free repair a daunting task. James Barrett Brown famously concluded that “the surgical repair of double cleft lips is about twice as difficult as in single clefts and the results are about half as good.”¹ This bleak view was largely the result of iatrogenic misadventures that are now well recognized and less commonly performed.

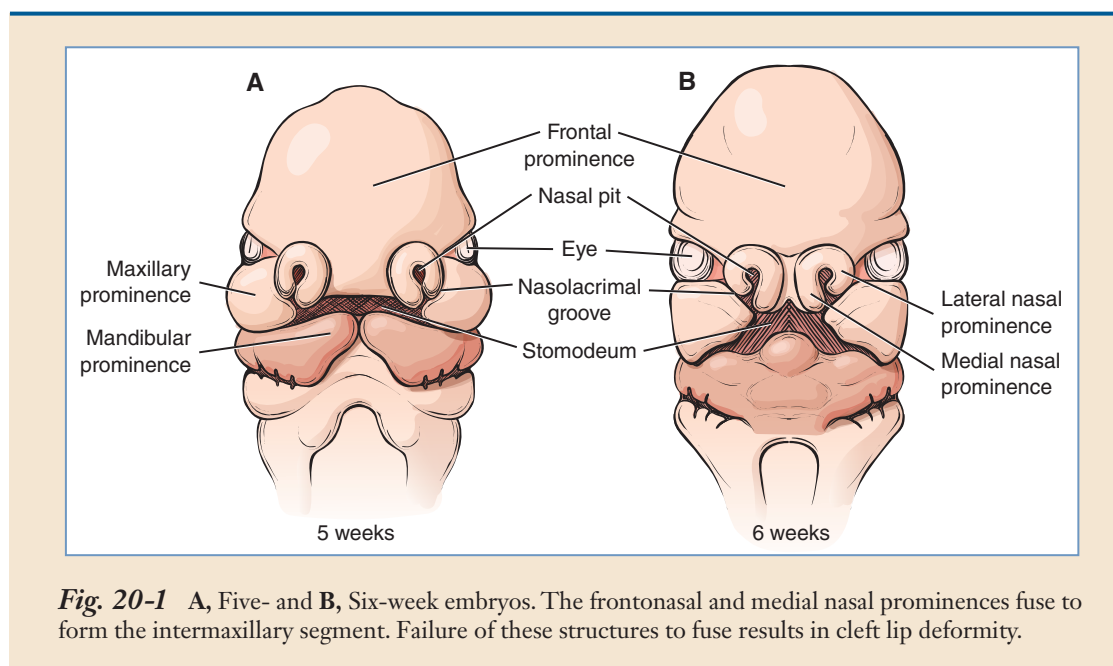
A classic example of resecting the protruding premaxilla or setting it back by resecting part of the vomer causes serious growth inhibition of the midface and markedly distorts the lip and nose.²⁻⁴ The use of banked forked flaps, which were used to elongate the columella in a delayed fashion, is another outmoded technique.⁵⁻⁷ Most surgeons now accept that the central white roll, vermilion, and mucosa are best reconstructed with tissue from the lateral lip elements and not the prolabium and that it is important to approximate the muscle from the lateral lip across the midline.⁸⁻¹⁰

ANATOMY

Normal Lip Embryology and Anatomy

Normal facial morphology results from the interaction of the five embryonic facial processes composed of neural crest and mesoderm-derived cells with an epithelial covering. The fronto-nasal and paired maxillary and mandibular processes undergo an intricate progression to fuse at the correct anatomic location.

The frontonasal process gives rise to the medial and lateral nasal processes, which form around the nasal placode. During week 6 to 7 of gestation, the medial nasal processes fuse and the maxillary processes merge to form the intermaxillary segment or the primary palate (Fig. 20-1).



The prolabium and premaxilla are formed from the frontonasal process, whereas the lateral lip elements and maxillary arches are derived from the maxillary processes.¹¹

The surface anatomy of the lip is divided into anatomic subunits consisting of the lateral lip elements, philtral columns, Cupid's bow, and median tubercle. The paired philtral columns separate the lateral lip elements from the philtrum. There is significant variation in the position of the philtral column at the lip-nose junction. Brooke et al¹² noted that most of the philtral columns originate lateral to the base of the columella rather than originating at the base of the columella. The double curvature of the lip resembles the bow of Cupid, the Roman god of love. There is significant variation in the depth of Cupid's bow between individuals, but all have paired convex peaks flanking a concave nadir. The median tubercle is a central area of fullness directly below Cupid's bow.

Three epithelial types distinguish the lip surface: (1) skin, (2) vermilion, and (3) mucosa. Each has distinct borders: the vermilion-cutaneous and vermilion-mucosal junctions. Mucosa and vermilion are completely separate entities, which are frequently incorrectly referred to as "dry vermilion" and "wet vermilion." Skin has stratified squamous epithelium and submucosal labial salivary glands. Vermilion is thinly keratinized stratified squamous epithelium without adnexal structures. Vermilion contains a high level of *eleidin*, a clear intracellular protein, which gives it the characteristic red color from the superficial blood vessels in the papillary layers.¹³ Mucosa is moist because of the salivary glands and is not normally exposed when the lips are in repose. The cutaneous roll or white roll consists of undulating, non-hair-bearing skin that separates the flat skin epithelium of the upper lip from the vermilion.

The orbicularis oris muscle forms a sphincter surrounding the upper and lower lips. The superficial component aids in facial animation and creates the philtral columns from decussating fibers inserting into the dermis. The deep component constricts the mouth to form a tight seal and puckers the lips.

Bilateral Cleft Embryology and Anatomy

In a complete bilateral cleft lip and palate (CL/P), the frontonasal process fails to fuse with the maxillary processes, creating a prolabium and premaxilla that are isolated from the lateral lip and maxillary arch elements.

The cleft prolabium is distinctive because of deficient soft tissues. The cutaneous portion lacks philtral columns. The normal vertical midline concavity is now convex. The cutaneous roll is hypoplastic and indistinct. The vermilion height of the prolabium is significantly reduced and hypoplastic compared with the lateral lip elements. A labial sulcus is generally absent. The prolabium does not contain any orbicularis oris muscle fibers. The premaxilla, no longer constrained by the maxillary arch elements, rotates anteriorly and elevates to an increasingly horizontal position.

The composition of the lateral lip elements is similar to the corresponding element in the unilateral cleft lip. Noordhoff's point¹⁴ describes the anatomic point on the lateral lip element where the vermilion-cutaneous and vermilion-mucosal junctions begin to converge. The cutaneous roll is well defined. Vermilion height is sufficient lateral to this point, but these structures become obscure and deficient medially. The orbicularis oris muscle fibers create a bulge as they abnormally insert into the alar base.¹⁵

The bilateral cleft nose deformity has the unique advantage of symmetry when compared with the unilateral cleft. The foundation of the deformity is based on a bilaterally hypoplastic maxilla. As the premaxilla protrudes, the lateral arch elements collapse, excluding the premaxilla from the arch. The bony derangement displaces the nostril bases laterally and posteriorly. The nasal tip broadens transversely and shortens sagittally. The lateral crura of the lower lateral cartilage lengthen and the medial crura shorten. This distortion results in medial displacement of the genua, effectively shortening the columella.¹⁶ As the lower lateral cartilages splay, the angle between the medial and lateral crura increases, which widens the soft triangle. The lower lateral cartilage and accessory cartilage complex attachment to the lateral piriform margin of the hypoplastic maxilla leads to posterolateral displacement and inward buckling of the ala and vestibular webbing.¹⁷ The abnormal insertion of the orbicularis oris muscle onto the alar bases contributes to their separation.

EPIDEMIOLOGY AND RISK FACTORS

The World Health Organization (WHO)¹⁸ recently reported the results from a study of an international database on orofacial clefts that collected reports from 54 registries in 30 countries. The population-based study included 7.5 million births and identified 7704 cases of cleft lip with or without cleft palate (CL $\pm$ P). The overall prevalence was 9.92 per 10,000 for CL $\pm$ P, 3.28 per 10,000 for CL, and 6.64 per 10,000 for CL/P. This was consistent with the prevalence of CL $\pm$ P in the United States (10.2 per 10,000), the United Kingdom (9.45 per 10,000), and Western Europe (11.88 per 10,000), as well as previously published regional studies.¹⁹⁻²¹ This was in contrast to the finding of 20.4 per 10,000 in Japan, although a previous regional study in Japan reported a prevalence of 13.4 per 10,000.²² In addition, 76.8% of CL $\pm$ P cases were isolated, 15.9% had malformations in other systems, and 7.3% occurred as part of a recognized syndrome. Of the registries recording laterality, 10.3% of CLs were bilateral and 30.2% of CL/Ps were bilateral.¹⁸ Multiple studies have noted that the prevalence of CL $\pm$ P is approximately 2:1 in males to females.^{23,24}

There is a higher risk of syndromic conditions or additional malformations in patients with bilateral clefts, and bilateral clefts occur more frequently in stillborn and aborted fetuses than in live births.²⁵

Nonsyndromic CL is thought to be multifactorial in origin, with a combination of genetic and environmental risk factors. Although the literature is replete with potential causes of clefts, there have been few clearly established links.

Maternal environmental exposure has been extensively investigated. Maternal tobacco use and excessive alcohol consumption in the first trimester have been associated with an increased risk of clefts in a dose-response relationship.²⁶⁻³⁶ Several studies have found a protective effect of periconceptional folic acid consumption and a reduced risk for CL $\pm$ P. However, the link is not as clear as it is for neural tube defects.³⁷⁻⁴⁰ Prescription medications with antifolate properties, corticosteroids, and anticonvulsants, such as carbamazepine, valproic acid, phenytoin, phenobarbital, and trimethadione, have been associated with an increased risk for CL $\pm$ P.⁴¹⁻⁴⁴ Prepregnancy diabetes mellitus, but not gestational diabetes, and obesity have recently been linked to increased odds ratios for CL $\pm$ P. The embryopathy is likely related to glucose intolerance and increased insulin resistance, but the exact mechanism has not been elucidated.⁴⁵⁻⁴⁸

PREOPERATIVE MANAGEMENT

Prenatal Diagnosis

The birth of a child is a life-changing event for any family and becomes of profound significance in the case of a facial anomaly. Since Christ and Meininger⁴⁹ initially reported on the detection of CL/P by ultrasonography in 1981, the initial presentation of a child with a cleft has drastically changed. In a recent systematic review of the literature, the detection rate of CL $\pm$ P ranged from 9% to 100% in a low-risk population without a false-positive diagnosis. However, several of these studies lacked adequate sample size power calculations and contained selection bias.⁵⁰ Furthermore, recent technological advancements in antenatal ultrasonography have improved the detection rate of CL $\pm$ P from 43% before 2007 to 86% after 2007.⁵¹ A similar detection rate of 81% was found in a prospective, nonselected, low-risk population of 35,924 women undergoing routine second-trimester prenatal ultrasonography in The Netherlands.⁵⁰

The prenatal diagnosis of a child with a cleft diagnosed by ultrasonography allows the surgeon to provide education, counseling, and support to the family, which can lessen the psychological impact at birth. Multiple studies have documented the positive effects of a prenatal diagnosis and that most families with an unsuspected child with a cleft would have preferred a prenatal diagnosis.⁵²⁻⁵⁴

There has been debate in the literature regarding the ethical issue raised by a prenatal diagnosis of a cleft, because it may increase the possibility of a termination of the pregnancy.⁵⁵⁻⁵⁹ Although there are case reports in the literature of pregnancy termination for isolated CL $\pm$ P, a recent study in The Netherlands did not find any difference in early detection and termination of a pregnancy.^{51,52}

Multidisciplinary Care

All children with clefts should be managed by a multidisciplinary team approach with input from specialists in surgery, pediatrics, genetics, otolaryngology, speech therapy, dentistry, orthodontics, audiology, social work, and a team coordinator. A pertinent history should include information on the pregnancy course, maternal exposure to teratogenic substances, delivery complications, prematurity, birth weight, and a family history of clefts. A special focus must be directed toward birth weight to ensure that the child is gaining weight. Feeding counseling, including the use

of modified feeding bottles such as the Haberman or Pigeon, should be provided. The physical examination should differentiate patients with nonsyndromic from syndromic cleft lip, with a specific look for defects, such as trisomy 13 and 18, Van der Woude syndrome, and 22q11 deletion syndrome.

INDICATIONS AND CONTRAINDICATIONS

Bilateral CL repair is indicated at any age if the child is healthy. A still widely accepted guideline is the “rule of tens,” in which the child is 10 weeks of age, weighs 10 pounds, and has a hemoglobin level of 10 g/dl. These criteria usually indicate that the child is healthy and that it is safe to proceed with surgery. A few practitioners advocate early repair, but very few repairs are carried out before 3 months of age. If presurgical orthodontics is used, the repair is delayed up to 6 months until the treatment is completed.

There are no contraindications to bilateral CL repair in a healthy infant. However, if there are any associated congenital anomalies or syndromes, the repair should be delayed until the other issues are addressed. Safety is of prime importance. There is no medical urgency to repair a bilateral CL.

PREOPERATIVE PLANNING

Presurgical Orthodontics

A skilled orthodontist is an integral part of a multidisciplinary cleft care team. However, debate continues regarding the appropriate age for orthodontic intervention. Proponents of presurgical orthodontia stress the importance of restraining the uncontrolled growth of the premaxilla while still allowing for the expansion of the lateral alveolar segments. The intent is to approximate a normal arch relationship to reduce the alveolar cleft widths and decrease tension on the CL repair.⁶⁰

Presurgical orthodontics can be implemented by active or passive mechanisms. The active Latham device requires application with the patient under anesthesia in the operating room. Two custom-fitted acrylic plates are fixated with pins to the palatal shelves and a transverse wire loop through the premaxilla. An intraoral ratcheted screw is turned daily to expand the lateral segments while an elastic chain retracts the premaxilla.⁶¹

The passive technique consists of alveolar molding with a custom fitted acrylic plate that is initiated within the first 2 to 3 weeks of life. The plate is positioned in place with elastics taped to the patient's cheeks and must be worn at all times except for cleaning. Every week the patient is evaluated, and the plate is modified to allow retraction of the premaxilla and alveolar displacement by shaving and adding acrylic. Over 2 to 3 months, the alveolar clefts are reduced to less than 5 mm, and nasal stents are added to the plate to provide an anterior force to reposition the lower lateral cartilages and lengthen the columella.⁶¹⁻⁶³

Bilateral Lip Adhesion

Bilateral lip adhesion is not routinely performed, but it has proponents who use it when presurgical orthodontics is not available or in patients with a severely protruding premaxilla. The goal of a lip adhesion is to temporarily close the clefts to create a constraining force against the protruding premaxilla. This is not an aesthetic procedure; thus it should not interfere with the

subsequent reconstruction. When planning a bilateral lip adhesion, the definitive lip repair should initially be marked, and all incisions must be made with these boundaries so that the important landmarks are not distorted and remain free of scar tissue. The most significant disadvantage of the bilateral lip adhesion is that there is no muscle reapproximation, which may lead to dehiscence of the adhesion.

Premaxillary Setback

Primary repositioning of the extremely protruding premaxilla has been described in the literature, but this should be abandoned, because there is significant risk for inducing midface growth restriction. If absolutely necessary, premaxillary setback should be performed secondarily in conjunction with an alveolar bone graft in mixed dentition or a segmental LeFort I osteotomy in skeletal maturity.

SURGICAL APPROACH

Principles

Too many infants born with bilateral cleft lip undergo old-fashioned, often multistaged, procedures, and later have to endure sundry revisions throughout childhood and adolescence. Despite the surgeon's efforts, the stigmata of the repaired bilateral cleft lip and nose remain painfully obvious—even at close distance. To the contrary . . . the appearance of a child with repaired bilateral cleft lip should be comparable to, and in many instances surpass, that of a repaired unilateral complete cleft lip.

—John B. Mulliken

Dr. Mulliken's statement⁶² is no longer viewed as overly optimistic. Several crucial principles have been elucidated as the surgical approach to the bilateral CL has evolved. By adhering to the following principles, iatrogenic deformities can be reduced and a better aesthetic result can be anticipated.

Simultaneous Repair to Maintain Symmetry

The primary advantage of the complete bilateral CL compared with the unilateral CL is symmetry. If the bilateral repair is staged, as was previously recommended to reduce tension, then the advantage is squandered, because it is more difficult subsequently to achieve symmetry. Furthermore, scar formation may not be identical at different ages. Asymmetries tend to become more pronounced as the child grows and will necessitate revisions.

Reconstruction of the Upper Gingivolabial Sulcus

The upper gingivolabial sulcus is deficient or nonexistent in the complete bilateral CL. If the sulcus is not reconstructed, the upper lip remains tethered to the alveolus and affects the aesthetic and functional result. The upper gingivolabial sulcus is reconstructed with the prolabial mucosa. The vermilion and mucosa are separated from the prolabial skin, draped over the premaxilla periosteum, and sutured to the periosteum near the nasal spine. This flap should be thinned to the thickness of a graft to decrease the bulk. Mucosal flaps from the lateral lip elements will be approximated in the midline to re-create the anterior wall of the sulcus.

Reconstruction of the Central Lip From the Lateral Lip Elements

The prolabial cutaneous roll lacks definition and should not be used to reconstruct Cupid's bow. The prolabial vermilion is hypoplastic and narrow and should not be used to reconstruct the central vermilion and dimple. With insufficient vermilion, mucosa becomes included in the repair. This mucosa is very obvious as a red, dry, and chapped patch where one normally sees vermilion.

The Cupid's bow should be reconstructed with cutaneous rolls and vermilion flaps from the lateral lip elements. These flaps will contain good quality tissue if the discrete anatomic point that marks the convergence of the vermilion-cutaneous and vermilion-mucosal junctions (Noordhoff's point¹⁴) is precisely chosen. Care must be taken when marking this point, because incisions made medially will create flaps with a hypoplastic cutaneous roll and vermilion with insufficient height to re-create a full median tubercle. Conversely, if the incisions are made too far laterally, then the transverse length of the lateral lip elements will be unnecessarily shortened, resulting in a tight upper lip. The cutaneous roll and vermilion flaps are sutured in the midline, and the caudal edge of the prolabial shield will set the depth of Cupid's bow.

Cutaneous Shield From the Prolabium

In keeping with the cleft tenet of "discard little to no tissue," several of the initial repairs preserved all of the prolabial skin, which resulted in a disproportionately wide central segment.⁶³ The prolabial skin that is included in the lip repair should be a relatively small, symmetrical shield.

Muscular Reapproximation

The aberrant insertions of the orbicularis oris muscles must be completely dissected free from the alar bases. Failure to do so will result in muscular bulges in the lateral lip elements. Midline reapproximation of the orbicularis, if at all possible, is important in reconstructing the natural sphincter, which is the most effective constraining force against a protruding premaxilla.

Primary Rhinoplasty

Initial efforts to address the bilateral cleft nasal deformity were centered on the thought that cleft rhinoplasty should be primarily deferred so as to not interfere with the growth of the nose. Another prevailing thought was that the columella was deficient and must be augmented with tissue, such as forked flaps recruited from the prolabium or nostril sills.⁶

Dissatisfaction with the stigmata that remained from these approaches grew until such pioneers as Broadbent and Woolf,⁸ McComb,⁶⁴ and Ortiz-Monasterio and Olmedo⁶⁵ challenged these concepts. The paradigm began to shift from secondary to primary nasal correction based on the principle that "the columella is in the nose,"⁶⁶ and the shortened columella was the result of malposition of the lower lateral cartilages. Furthermore, primary cleft rhinoplasty was shown not to impair facial growth.^{17,65,67}

A brief review of the approaches to primary rhinoplasty will help to elucidate the contentious issues. McComb⁶⁸ described a "flying bird" incision across the lower aspect of the nasal tip, which continued bilaterally as alar rim incisions. The author thought that the accurate reapproximation of the alar cartilages, reestablishment of the columella, and narrowing of the nasal tip with a V-Y closure justified the creation of an external nasal scar⁶⁸ (Fig. 20-2).

Mulliken⁶⁹ championed bilateral alar rim incisions to access the middle crura and genua of the lower lateral cartilages. The domes are then approximated with an interdomal mattress suture. The lateral crura are also suspended to the ipsilateral upper lateral cartilages. Skin is excised from the soft triangle and lateral columella to narrow the tip, define the columellar-lobular junction, elongate the nostrils, and narrow the columellar waist⁶⁹ (Fig. 20-3).

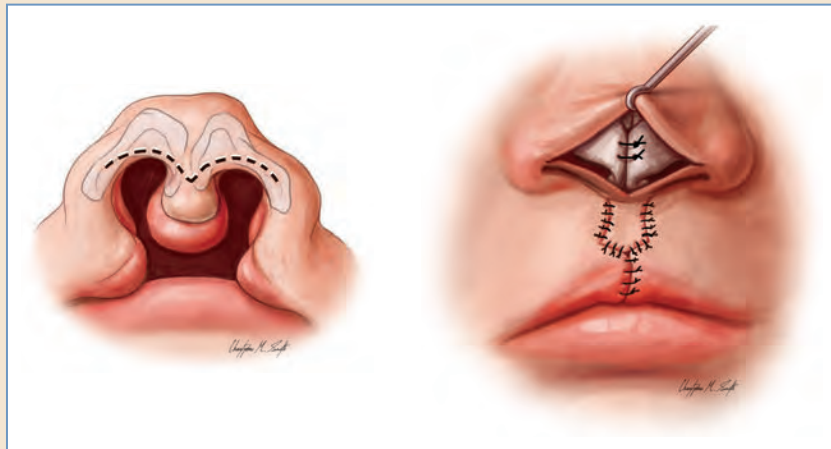


Fig. 20-2 McComb's original "flying bird" technique for primary cleft rhinoplasty.

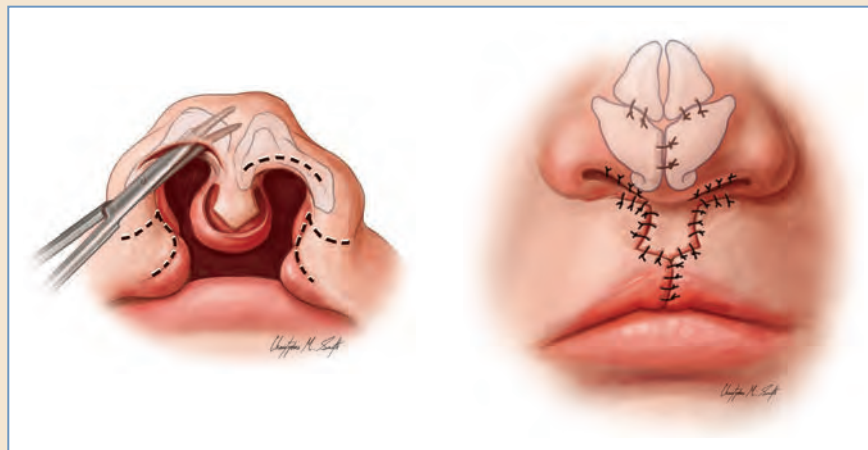


Fig. 20-3 Mulliken's technique for primary bilateral cleft rhinoplasty.

Trott and Mohan⁷⁰ extended bilateral alar rim incisions continuously with the prolabial shield incisions. The prolabium, columella, and nasolabial crease are then elevated as a single unit to gain open access to the nasal tip. Caution should be used with this approach, because the blood supply to the distal tip of the prolabium is based on the dorsal nasal skin and is at risk for ischemia. Cutting's approach improved the vascularity of the prolabial-columellar flap by elevating it at the membranous septum.⁷¹

Despite the more aggressive approach to a primary rhinoplasty, most patients will still require a secondary rhinoplasty to obtain an optimal result. Many authors correctly point out the long-term negative effects of poorly placed incisions on the skin that produce excessive scars. Incisions used for alar rim excisions, lateral rhinotomies, nasal tip access and resections, and primary columellar lengthening can cause secondary CL nasal deformities that are more difficult to correct than the natural deformity itself.

These authors argue that a conservative primary rhinoplasty should be used to limit iatrogenic deformities. Another reason to delay the definitive rhinoplasty is because the lower lateral cartilages are partially deformed as a result of maxillary hypoplasia at the lateral piriform margins and that to re-create the nasal tip and columella, the lower lateral cartilages must be anatomically repositioned on a corrected skeletal base. This is achieved only after appropriate alveolar bone grafting, orthodontics, and where necessary, orthognathic surgery. Performing a cleft rhinoplasty before these procedures may artificially alter the final aesthetic landmarks.⁷²

SURGICAL MARKINGS

The surgical markings and subsequent description of the technique have slight modifications to the Fisher technique. The markings and techniques of Fisher,¹⁶ Mulliken,⁶⁹ and Chen and Noordhoff¹⁴ are illustrated to highlight the similarities and differences (Figs. 20-4 through 20-6).

The essential objective of obtaining symmetry in a bilateral cleft lip repair starts with accurate preoperative marking.

A fine-tipped pen, either a sharpened Q-tip or fountain pen, is used to make the initial marks, which are then tattooed with a 25-gauge needle and dye. Gentian violet or brilliant green dyes are better options than the more commonly used methylene blue, which tends to “run.”

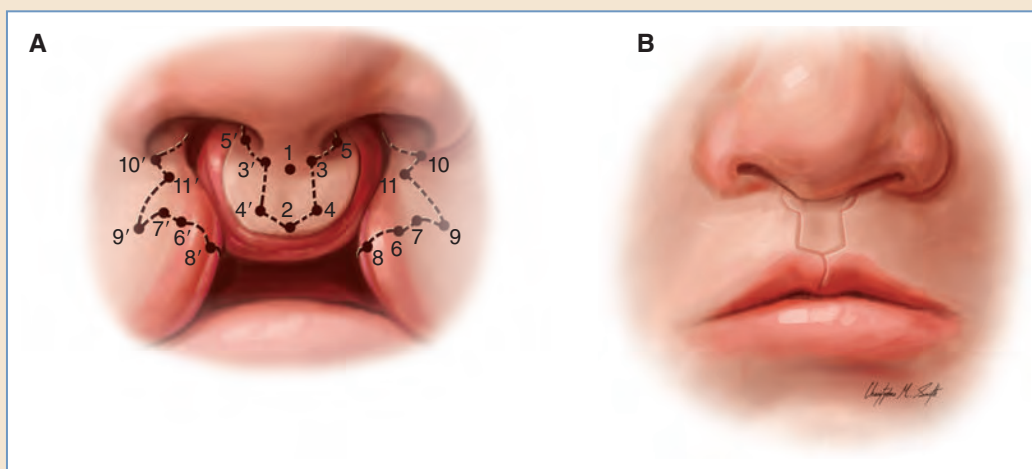


Fig. 20-4 Fisher's markings and technique. **A**, Point 1 marks the midline of the columella. Point 2 marks the midpoint of the prolabium, just above the deficient vermilion-cutaneous junction. Points 3 and 3' at the columellar base and 4 and 4' at the caudal end mark the outline of the tie-shaped prolabial skin. Point 5 is just lateral to the lip-columellar crease. Point 6 indicates the position of Noordhoff's point. Point 7 is marked just lateral to and above the cutaneous roll. Point 8 is below Noordhoff's point on the vermilion-mucosal junction. Points 6, 7, and 8 form a straight line perpendicular to the vermilion-cutaneous junction. Point 9 is marked just above the cutaneous roll lateral to point 7. The distance between points 7 and 9 should equal the distance between points 2 and 4. Point 10 is placed medial to the alar insertion and meets point 5 at the height of the lip repair at the anterior nostril sill. Point 11 is positioned near the cleft margin at a distance from point 9 that is equal to the distance between points 3 and 4 and at a distance from point 10 that is equal to the distance between points 3 and 5. **B**, Scar outline of a completed Fisher repair.

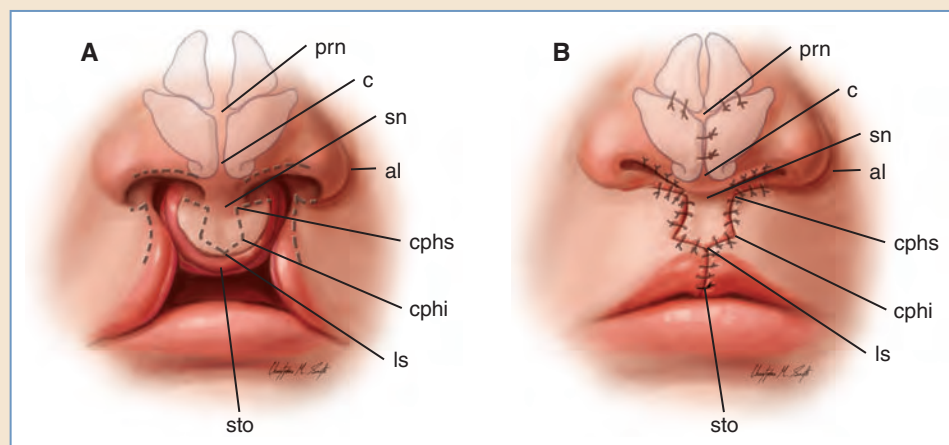


Fig. 20-5 Mulliken's markings and technique. **A**, Mulliken's technique uses anthropometric terms for the key points. The similarity to Fisher's points is readily apparent. **B**, Completed bilateral complete Mulliken cleft lip repair and primary rhinoplasty. (*prn*, Pronasale; *c*, highest point of columella nasi; *sn*, subnasale; *al*, ala nasi; *cphs*, crista philtri superior; *cphi*, crista philtri inferior; *ls*, labiale superius; *sto*, stomion.)

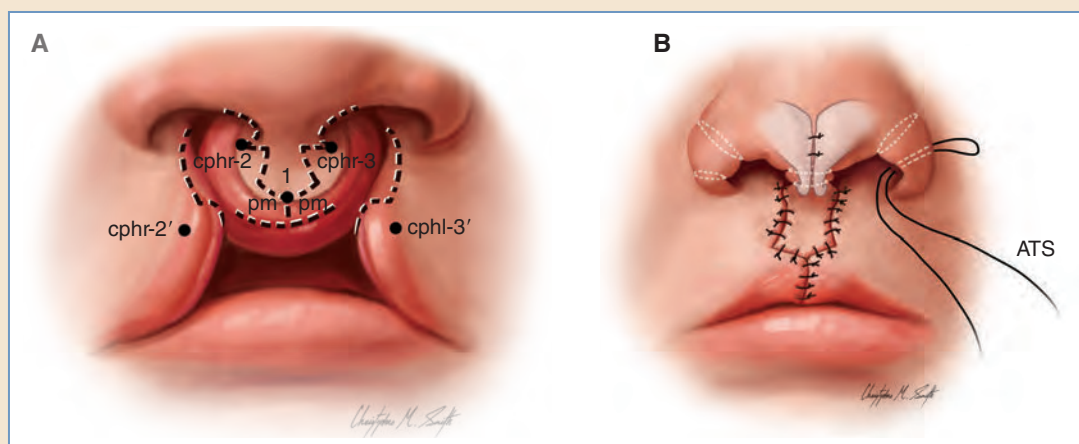


Fig. 20-6 **A**, Surgical marking after Noordhoff. Point 1 is the central point of the Cupid's bow. The lateral points of the Cupid's bow, on the right side (*cphr-2*), and on the left side (*cphr-3*) keep the total width of the Cupid's bow a maximum distance of 4 to 5 mm. The vertical limbs extend to the base of the columella, with the base narrowed to 3 to 4 mm wide. Points *cphr-2'* and *cphi-3'* are the anatomic points for the base of the philtral column on the lateral lip. These points are 3 to 4 mm lateral to the converging junction of vermilion and mucosa (red line) and the cutaneous roll. This point is where the vermilion first becomes its widest and is known as Noordhoff's point. The prolabial mucosa (*pm*) is used for reconstruction of the nostril floor. The central part of the prolabial mucosa is used for lining the premaxilla. **B**, The skin incisions are closed with fine absorbable sutures. Alar transfixion sutures (*ATS*) are placed to accentuate the ala-facial groove, support the lower lateral cartilage, and minimize vestibular webbing.

The surgeon should take the patient's race and parent's appearance into consideration when marking the philtral flap, but in general it is marked as a tie-shaped shield. Some authors have recommended that the shield width not be wider than 4 mm because of the widening that occurs as the child grows.^{73,74} This is probably less important than ensuring that the shield is symmetrical. The midpoint of the lip columella junction (1) and the midpoint of the prolabium in the normal skin just above the defective cutaneous white roll (2) are marked first. The width of the apex of the cutaneous shield is set by marking points 1.5 to 2 mm lateral to the central point in the lip-columellar junction (3,3'). Marks are also placed superiorly around the base of the columella (5,5').

The width inferiorly is set by making marks approximately 2.5 mm superolaterally on either side of the central point (4,4'). These points must be made cephalad to the deficient cutaneous roll of the prolabium and will become the peaks of Cupid's bow. The position of these two points will also determine the shape of Cupid's bow. As previously mentioned, the prolabial cutaneous roll and vermilion are of deficient quality and should not be included in the repair.

The markings for the lateral lip element begin at Noordhoff's point. This is the anatomic landmark at the convergence of the vermilion-cutaneous junction and the vermilion-mucosal junction with a full cutaneous roll (6,6'). This point must be accurately marked as shifting laterally and will include a full cutaneous roll and vermilion of sufficient height but will horizontally shorten the lateral lip. Conversely, shifting the point medially will include an ill-defined cutaneous roll and vertically deficient vermilion. Additional points are marked above the cutaneous roll at Noordhoff's point (7,7') and at the vermilion-mucosal junction below Noordhoff's point (8,8'). These points form a straight line perpendicular to the lateral lip element.

The location of the remaining points will depend on the prolabial markings. Another point is placed above the cutaneous roll lateral to the mark at the edge such that the length will match the midline to the superolateral points of the prolabial skin (9,9'). This will reconstruct the cutaneous roll above Cupid's bow.

A point located near the cleft margin such that this line is the same length as the lateral edge of the prolabial skin shield sets the vertical height of the lateral lip element (10,10'). Approximating the tissue along the cutaneous-mucosal junction, which is clearly seen intranasally along the anterior septum and nostril base, will close the nostril sill and anterior nasal floor (Box 20-1; see also Fig. 20-4, A).

Box 20-1 Bilateral Cleft Lip Markings

1. Midline of the lip-columellar junction
2. Midline cephalad to the prolabial cutaneous roll
3. Base of the philtral flap approximately 1.5 mm lateral to point 1 in the lip-columellar crease
4. Proposed peak of Cupid's bow approximately 2.5 mm superolateral to point 2
5. Nostril sill
6. Noordhoff's point
7. Cutaneous roll above Noordhoff's point
8. Vermilion-mucosal junction below Noordhoff's point
9. Lateral to point 7 above the cutaneous roll keeping lines 7-9 equidistant to lines 2-4
10. Vertical height of the lateral lip element keeping lines 9-10 equidistant to lines 3-4
11. Proposed closure of nostril sill keeping lines 10-11 equidistant to lines 3-5

SURGICAL TECHNIQUE

Bilateral infraorbital nerve blocks are performed with a 50:50 mix of 0.25% bupivacaine and 1% lidocaine with 1:100,000 epinephrine. In addition, the lateral lip elements, upper buccal sulcus, prolabium, alar bases, and inferior turbinates are infiltrated with local anesthetic.

After adequate time is allowed for the analgesic and vasoconstrictive properties to take effect, dissection is started on the prolabium.

An acute angle blade, such as a No. 11 blade scalpel, is used to incise the tie-shaped prolabial skin shield. When available we favor the use of a 300 disposable ophthalmic microknife. The incision is extended laterally around the base of the columella and then posteriorly around the premaxilla-septal junction, along the line of the skin and mucosa of the anterior nostril, which marks the medial edge of the cleft. A superiorly based mucoperichondrial flap is raised from the anterior septum for later closure of the nasal floor.

The prolabial shield is then cephalically elevated in a subcutaneous plane to the base of the anterior nasal spine. The skin lateral to the shield is deepithelialized to add bulk for the reconstruction of the philtral columns. A prolabial mucosal flap is caudally elevated in a submucosal plane. The flap should be thinned to the thickness of a mucosal graft. The mucosal flap is inset over the anterior premaxilla with a 5-0 absorbable horizontal mattress suture to the premaxillary periosteum to reconstruct the posterior wall of the gingivobuccal sulcus (gingival surface of the prolabial alveolus) (Fig. 20-7).

The lateral lip elements are addressed first with a full-thickness, perpendicular incision to the lateral lip element starting at the cutaneous roll above Noordhoff's point (point 7) and then continuing perpendicularly through the vermilion and mucosa of the inferior aspect of the lip (lines 7 and 8).

An incision is now made from the lateral point above the cutaneous roll to the nostril base. The incision continues superiorly and posteriorly along the junction of the cleft margin and nasal vestibular mucosa at the base of the inferior turbinate. A superiorly based mucoperiosteal flap can be raised from the inferior turbinate to help with closure of the nasal floor. A strip of the cleft margin is excised down to the convergence of the CL, alveolus, and vestibule. The excised cleft tissue is discarded.

An upper buccal sulcus incision is made laterally from the cleft margin. The alar base is freed from the piriform rim in a supraperiosteal plane. This dissection needs to be extended laterally and superiorly enough to allow complete mobilization of the soft tissue of the lateral lip element and the alar base to close without tension. The extent of the dissection will often extend to the infraorbital nerve, and care should be taken not to injure the nerve.

The orbicularis oris muscle is freed from the abnormal attachment to the piriform rim as part of the dissection releasing the alar base. It is also freed from the alar base using scissors to dissect from the labial and nasal layer up to the subdermis of the alar groove. The remaining dissection separates the orbicularis from the skin, mucosa, and vermilion.

Closure starts by suturing the nasal floor with a 5-0 suture on a small semicircular needle, such as a G-2 needle. If needed, a superiorly based inferior turbinate flap is used to close the posterior part of the nasal floor, suturing the inferior turbinate flap to the septal mucoperichondrial flap. Suturing the junction of the skin-mucosal junction from the alar base laterally and the septum medially aligns the anterior part of the nasal floor closure.

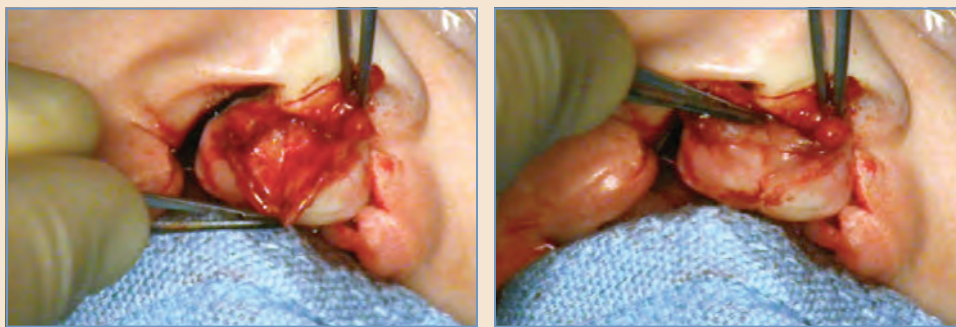


Fig. 20-7 Creation of the labial sulcus. The prolabial mucosa is thinned and then sutured near the base of the columella to line the prolabial alveolus.

Mucosa from both lateral lip elements is advanced to the midline and approximated with 4-0 chromic sutures. The superior aspect of the lateral lip mucosa is anchored to the premaxilla periosteum to set the depth of the gingivobuccal sulcus.

The lateral lip vermilion-mucosal junctions are approximated with a 4-0 chromic suture. The muscle approximation starts with an interrupted suture in the pars marginalis of the muscle that has been dissected from the vermilion and mucosa. This suture is left long, and a downward tension on this suture facilitates the progressive approximation of the muscle with buried 4-0 absorbable sutures.

The vermilion, skin, and nostril sill are approximated with interrupted dermal 5-0 absorbable sutures. Final closure is then achieved with interrupted 6-0 rapidly absorbing sutures or even cyanoacrylate glue if the edges are well approximated.

We currently perform a McComb type of primary cleft rhinoplasty.⁶⁷ The lower lateral cartilages are freed from the overlying nasal skin by dissecting from the open wound at the nostril bases and the base of the columella. The dissection is carried over the domes of the lower lateral cartilages, and the tissue is freed from between the two medial crura. The dissection is carried up onto the nasal dorsum and above the upper lateral cartilages. Alar transfixion sutures are then placed with the use of an absorbable monofilament such as 4-0 PDS to approximate and elevate the domes of the lower lateral cartilages. These sutures are tied intranasally and have the effect of refining the nasal tip and lengthening the columella. Additional bolster sutures help shape and contour the nostril sidewalls and bases as illustrated in Fig. 20-6, *B*.

POSTOPERATIVE CARE

Patients are allowed to resume oral intake with the same bottle they used preoperatively or the breast in cases of isolated cleft lips. Intravenous fluids are continued until adequate oral intake is achieved. Patients are discharged the same day as surgery with postoperative instructions regarding incision care and a limited course of liquid narcotic pain medication. The vast majority of patients have transitioned to only acetaminophen for pain control by postoperative day 1. All cutaneous sutures are absorbable and do not require removal.

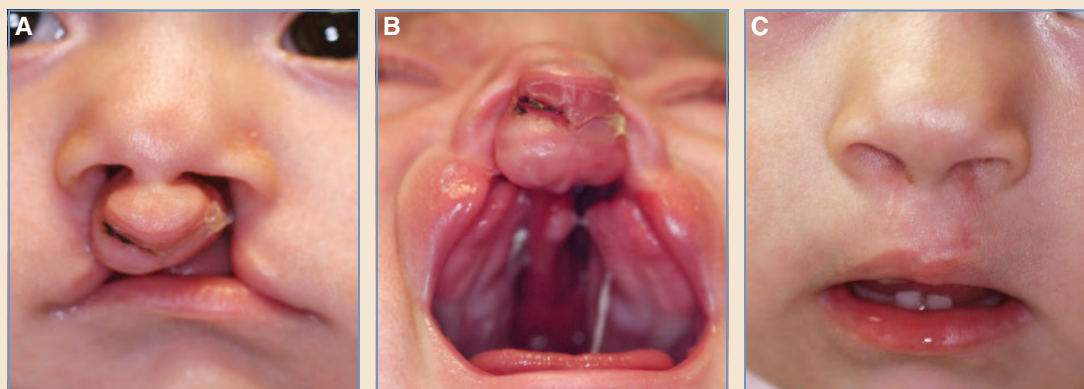


Fig. 20-8 A and B, Preoperative bilateral CL/P with prominent prolabial segment. C, One year after surgery.

Arm restraints are not used in our patients. In addition to being irritating to the patient, splints do not decrease complications such as dehiscence or fistulas when compared with non-restrained patients. The children very quickly grasp that interfering with the new repair hurts and instinctively protect themselves.⁷⁵ Follow-up is arranged within 2 weeks, at which time the parents are instructed on scar massage. A clinical example is shown in Fig. 20-8.

COMPLICATIONS

Although infants are obligate nasal breathers, this is generally not an issue at 3 months of age, and airway obstructions are exceedingly rare. Anesthetic complications are also rare and can be minimized by including a pediatric anesthesiologist in the multidisciplinary care. Bleeding, infection, and dehiscence are infrequent and can be managed with meticulous hemostasis and local wound care. Late scar revisions are the most frequently required, but true hypertrophic scars are uncommon.

Incomplete Bilateral Cleft Lips

Bilateral CL can present as a broad spectrum encompassing symmetrical complete, symmetrical incomplete, and asymmetrical variants. An asymmetrical bilateral CL may be complete or incomplete, with a contralateral incomplete or lesser form cleft (minor, microform, or mini-microform).⁷⁶

Symmetrical incomplete bilateral clefts may require additional tissue resection because of the long lateral lip elements. These resections should be placed medial to the alar base. The muscle closure is approached in the same way as a complete cleft, and the vermilion reconstruction should rely on the lateral lip elements. The buccal sulcus and nasal floor do not need to be addressed.

Asymmetrical clefts are challenging and must be repaired asymmetrically, with additional tissue resected medial to the alar base on the incomplete side. Asymmetrical clefts with a lesser form pose a challenge regarding the extent of the repair and will depend on whether the lesser

form even requires a correction. If the deformity is minimal, then the contralateral side can be repaired as a unilateral cleft. If the deformity is significant, then the repair should proceed as an asymmetrical bilateral CL.

Secondary Cleft Rhinoplasty

Despite the trend to perform more aggressive primary cleft rhinoplasties, secondary cleft rhinoplasties are still frequently necessary. Ideally, the secondary cleft rhinoplasty is performed after skeletal maturity and any orthognathic surgery. However, if the deformity is severe or the child is suffering from any psychological stress, such as teasing, an earlier correction should be undertaken.

Secondary rhinoplasties to correct bilateral clefts usually have to deal with a short columella, a wide poorly defined nasal tip, and splayed nostril bases. Correction requires a combination of an open approach, extensive mobilization and repositioning of the lower lateral cartilages, nasal osteotomies, and extensive use of cartilage grafts.

CONCLUSION

A bilateral CL/P represents the extreme form of clefting and a real challenge to the family and team taking care of the child. Improvements in high-resolution ultrasound technology have increased the frequency of prenatal detection and diagnosis, which in turn has allowed us to do a better job of preparing parents for the birth and subsequent treatment. Advances in nasoskeletal molding have also improved the initial outcomes and certainly improved the outcomes when addressing the vexing problem of a short columella.

KEY POINTS

- All children with clefts should be managed by a multidisciplinary team, with initial counseling on feeding techniques.
- Premaxillary setback will cause serious midface growth restriction.
- A bilateral CL should be repaired simultaneously to preserve symmetry.
- The cleft prolabium contains several deficiencies. The cutaneous roll and vermilion height are hypoplastic, and there is an absence of orbicularis oris fibers or philtral columns.
- Cupid's bow should be reconstructed with cutaneous rolls and vermilion flaps from the lateral lip elements.
- Preservation of all of the prolabial skin will result in a disproportionately wide central segment. The prolabial skin should be used to create a small, symmetrical, cutaneous shield, with which to form the philtral columns.

Continued

KEY POINTS (continued)

- The upper gingivolabial sulcus is deficient or nonexistent. A caudally based prolabial mucosal flap is draped over the premaxilla to reconstruct the posterior border of the sulcus. Mucosa from the lateral lip element will reconstruct the anterior border of the sulcus.
- The orbicularis oris muscle must be completely freed from the abnormal insertions on the alar bases and lateral lip skin. Reapproximation in the midline provides a constraining force against the protruding premaxilla.
- The nostril floor can be closed with a superiorly based mucoperiosteal flap based on the inferior turbinate and septum.
- The shortened columella is caused by malposition of the lower lateral cartilages. Banked forked flaps should be abandoned and not used to elongate the columella. Primary cleft rhinoplasty has not been shown to impair facial growth.

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21

Secondary Deformities of the Cleft Lip and Nose

Jeremy P. Warner • Bruce S. Bauer



Most cleft lip patients seen for revision have relatively minor deformities, because the principles described in Chapters 19 and 21 were followed for the primary repair, with attention to the critical landmarks and meticulous technique. A cleft lip nasal deformity may be the most visible evidence of a prior cleft. Busy cleft surgeons see patients whose lip repairs were carried out by less skilled surgeons and whose complications of healing and scar management have resulted in quite significant deformity. Revisions range from minor realignment of landmarks with direct excision or Z-plasty to major revisions involving full-thickness layered repair or Abbé flap reconstruction. This chapter presents a review of the more common minor revision procedures and a discussion of the principles of major lip revision. The main focus is an in-depth discussion of the treatment of secondary deformities of the cleft lip nose.

We think that a thorough understanding of the nasal deformities seen and the procedures required to correct them best begins with a brief discussion of the primary deformities of unilateral and bilateral clefts. Surgeons' opinions vary regarding the appropriate degree of primary correction and how much of the deformity remains for secondary correction. Most surgeons who routinely perform cleft lip repair agree that an uncorrected nasal deformity will belie even the best lip repair and invisible scar. Excessive dissection in a primary nasal repair may affect long-term nasal growth. Surgeons respect this to various degrees depending on their comfort level. Therefore the spectrum of secondary nasal deformities varies from minor asymmetries to marked asymmetry, hypoplastic growth, and airway obstruction at skeletal maturity.

SECONDARY LIP DEFORMITIES

Isolated secondary lip deformities can develop in an otherwise well-performed lip repair and can be corrected by minor revisions. When a primary repair was not performed optimally, these deformities may not be isolated and are better addressed by a major revision. Lip shape, symmetry, movement, and associated nasal symmetry are dependent not only on alignment of the soft tissue landmarks, but also on the bony support beneath the lip and nose. Proper release of the soft tissues during a repair may provide excellent symmetry even in the presence of underlying maxillary hypoplasia, but failure to fully release the lip and nasal structures can result in significant collapse of the soft tissue with increased deformity even when the lip has been well repaired otherwise. Most lip revision procedures are relatively straightforward, whereas others defy simple explanation. For these cases, surgeons need to “separate the parts” and occasionally rely on basic plastic surgical ingenuity to place them in the final symmetrical position (Fig. 21-1).



Fig. 21-1 **A**, Relatively minor asymmetry with fullness of the vermilion and mucosa, symmetrical lip length and shape of the Cupid's bow, and drift of the alar base. **B**, Symmetry of the lip and nose is overshadowed by the significant collapse of the alar base and heminose, which resulted from a pronounced deficiency of the underlying bone. **C**, Although the nose is relatively symmetrical, the repair did little to align the lip landmarks or correct the underlying muscle. What remains is similar to an occult cleft. **D**, Initially, this appeared to be a slightly short cleft side with a displaced columella. It was actually related to the whole lip being oriented off midline, with the columella vertically in line with the high point of the Cupid's bow. **E**, In this case, the deformity of the lip and nose are from an atypical shift of the columella to overlie the normal peak of the Cupid's bow. This may have happened secondary to early overresection in the nostril floor on the noncleft side. The lip is elongated on the normal side to a greater degree than it is short on the cleft side. **F**, This bilateral repair is similar to a lip adhesion, because it lacks a muscle repair and alignment of the vermilion-cutaneous or vermilion-mucosal junction to augment the central tubercle.

Minor Revisions of the Lip and Nose

Alar Base

Alar base asymmetry is a universal feature of primary cleft deformity and is not always in proportion to the degree of lip clefting.¹ Because skeletal deficiency is generally not corrected at a primary repair, some degree of residual alar base asymmetry in the AP dimension is frequently present in the secondary deformity. The repositioned alar base tends to drift posteriorly after repair, reflecting the severity of the original and uncorrected skeletal base asymmetry (cleft-side maxillary hypoplasia)² (see Fig. 21-1, *A* and *B*). Both the soft tissues and the underlying skeleton need to be considered before attempting to correct this problem; therefore the timing should be decided based on the degree of underlying skeletal support. The debate regarding the pros and cons of primary bone grafting is discussed in Chapter 28. However, early placement of a primary graft may have the advantage of improving the skeletal support.^{3,4} The choice of procedure for correcting the drift of the alar base depends on the direction of distortion.

In cases in which the alar base position is significantly affected by the underlying bony hypoplasia (see Fig. 21-1, *B*), it is best to wait until after an alveolar bone graft and graft to the piriform margin. If the vertical height of the lip is in balance, with the peak of the Cupid's bow on the normal and cleft side symmetrical, minor asymmetries of the alar bases in the vertical dimension can be addressed by a Z-plasty. If the alar insertion is too low, reciprocal transposition of the alar base with a triangle taken from cephalad within the nostril sill and vestibule can be helpful; however, care must be taken not to introduce skin of different character (thickness, color, hair, and scar) onto the upper lip (Fig. 21-2, *A*). If the alar insertion is too high, reciprocal transposition of the alar base with a triangle taken from the upper lip can be performed (Fig. 21-2, *B*). In general, if the transverse wedge resection is confined to the skin and subcutaneous tissue, the likelihood of elevating the lip on that side is minimal (Fig. 21-2, *C*). (A full-thickness excision,

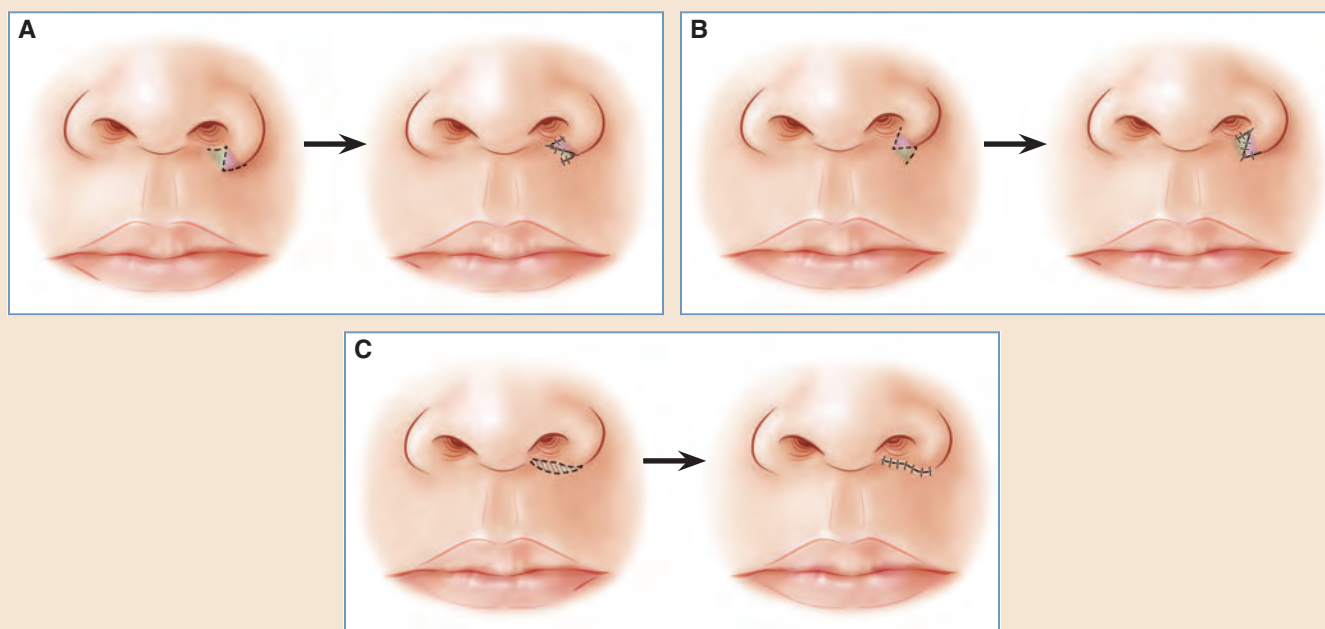


Fig. 21-2 Alar base repositioning. **A**, Z-plasty to elevate an alar insertion. **B**, Z-plasty to lower an alar insertion. **C**, An elliptical excision.

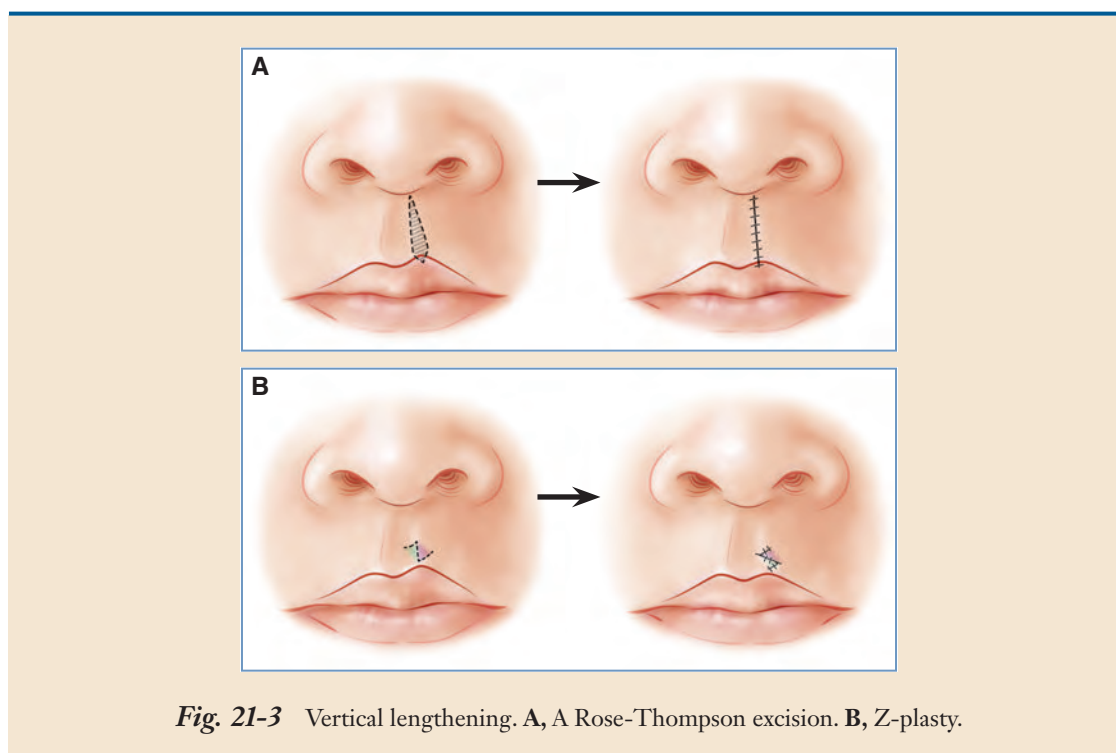


Fig. 21-3 Vertical lengthening. **A**, A Rose-Thompson excision. **B**, Z-plasty.

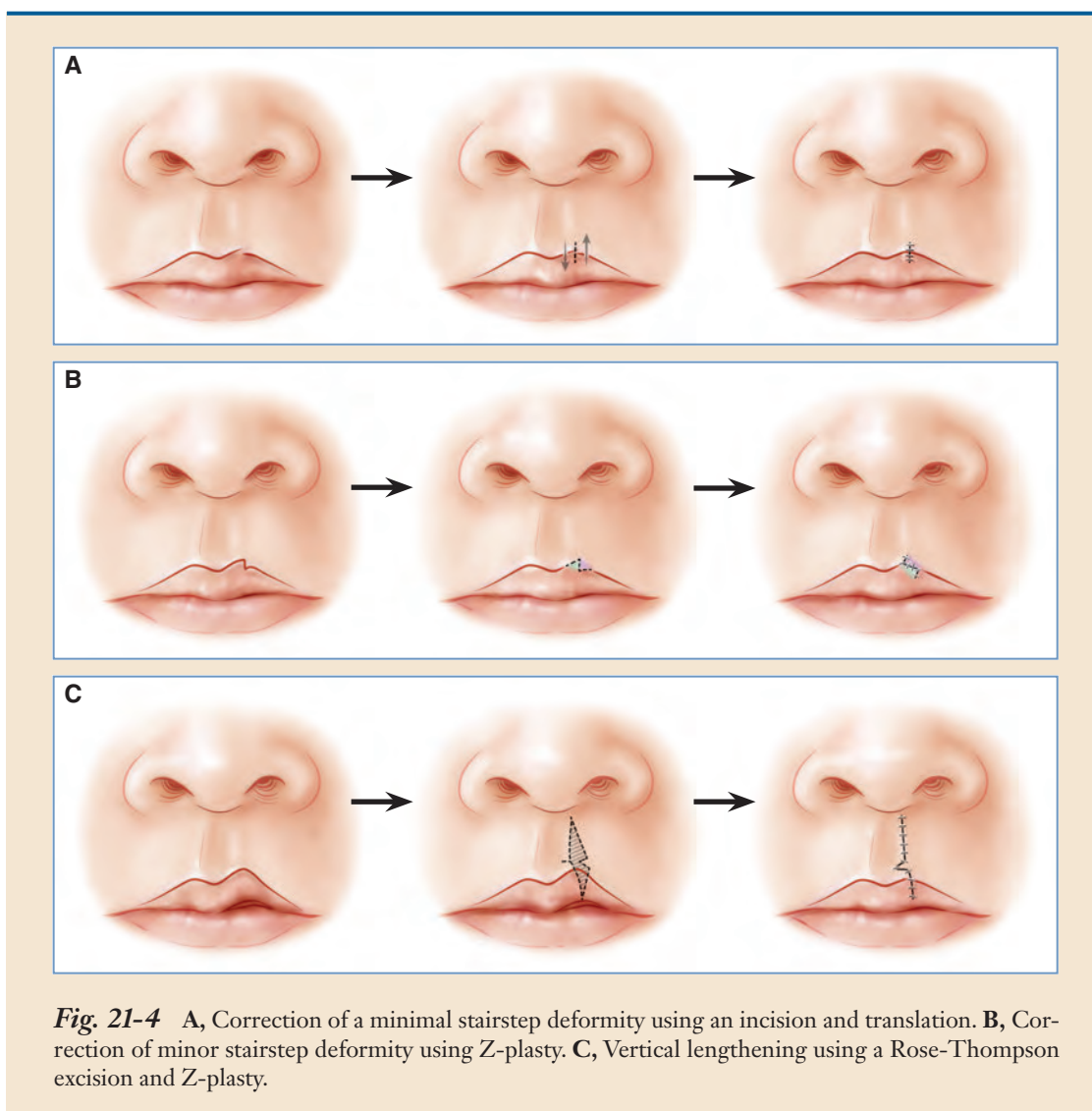
including muscle, with fixation sutures along the piriform margin may not fully correct a vertically long lip.) For patients who have already had a graft and have good skeletal balance without the need for a maxillary advancement, a triangular-shaped, composite graft of conchal cartilage with attached postauricular fascia and fat creates an excellent, stable additional platform to elevate the alar base. The graft can be set through an incision in the nostril sill or along the medial edge of the alar base.

Nostril Sill

The nostril sill may be inappropriate in width, depressed, or excessively scarred. A broad sill can be reduced fairly easily with an elliptical excision. Great care is needed during the primary procedure to preserve the nostril sill to prevent a constriction of the nares. A sill that is too narrow can be augmented with skin from the upper lip (see Fig. 21-2, *A*) or lateral to the alar crease (see Fig. 21-2, *B*), but not without introducing a donor-site scar. A depressed nostril sill is rarely seen in isolation. It can result from an incomplete muscle repair and present with a lateral lip muscular bulge. Treatment is therefore done in concert with excision of the cutaneous lip scar, including scar through the area of muscle dehiscence, and muscle lengthening with interdigitating muscle bundles (Figs. 21-3, *A*, and 21-4, *A* and *B*). As part of a major lip revision, this maneuver is more likely to lengthen a shortened lip (and effect a lowering of the peak of the Cupid's bow) than a small Z-plasty (as shown in Figs. 21-3, *B*, and 21-4, *A* and *B*). Therefore it is generally best treated by a major revision.

Cutaneous Lip

As with scar revision, revising the cutaneous component of a lip repair scar should be considered if it can improve the scar appearance. However, a major revision offers an opportunity to repo-



sition a scar to better mirror the philtral column on the noncleft side. For this reason, it may be preferred over a minor revision. Although past dogma suggested that the best lip scar resulted from early repair, meticulous closure and active massage in the months after surgery probably produces a better scar. Even a new scar will be less visible if overall lip symmetry is improved.

Minor vertical height deficiencies (up to 1 mm) from underrotation of the cleft-side peak of the Cupid's bow can be corrected using a Rose-Thompson excision and closure^{5,6} (see Fig. 21-4, A). Moderate vertical height deficiencies greater than 1 mm can be corrected with a Z-plasty. The lower portion of the lip scar is opened or elliptically excised, with or without excision of the upper lip scar as indicated, and a small opening incision is made above the white roll in the medial lip element to adequately lower the cleft-side peak of the Cupid's bow. The resultant defect is then filled with a triangular flap equal in size to the defect pared from the lateral lip element (see Fig. 21-4, B). If a Rose-Thompson lengthening is incorporated into the design, the triangular flap need not be large (see Fig. 21-4, C).

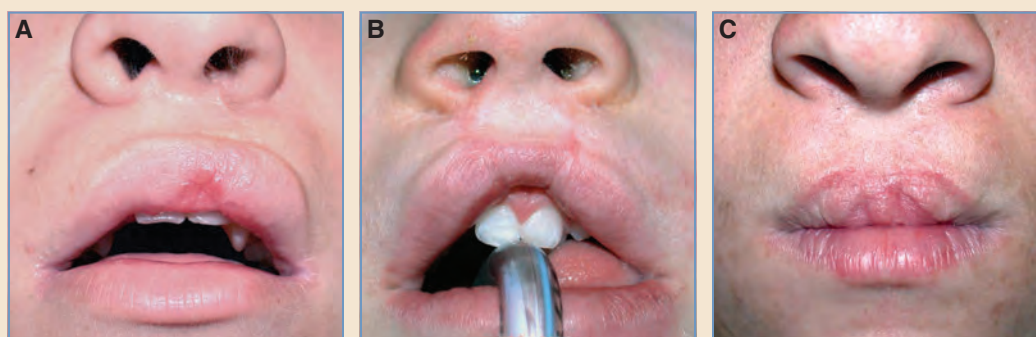


Fig. 21-5 A, A unilateral cleft lip with a stairstep deformity of the vermilion-mucosal junction (red line) and central vermilion deficiency. B, A bilateral cleft lip. Retention of the prolabial cutaneous roll and the vermilion has resulted in a Cupid's bow of poor quality and a cutaneous roll and vermilion of inadequate vertical height. C, A bilateral cleft lip. In this case, the lateral lip elements have been pared medial to Noordhoff's point.⁷ A poor-quality cutaneous roll and a vermilion of inadequate vertical height are retained. Mucosa is exposed below the central vermilion.

Significant vertical height discrepancies often occur in combination with other deformities and are usually best managed by a major revision.

Vermilion-Cutaneous Junction

The initial repair is the best time to correctly align the vermilion-cutaneous and vermilion-mucosal junctions. As emphasized in Chapters 19 and 20, correctly choosing Noordhoff's point and correcting a medial vermilion deficiency are critical. Discrepancies are corrected using standard maneuvers to match adjacent flap lengths.

Small stairstep deformities (1 mm or less) of the vermilion-cutaneous junction can be corrected with a vertical incision through the scar and translation of the medial and lateral lip elements (Fig. 21-5, A). Larger stairstep deformities (greater than 1 mm) may be corrected with a Z-plasty, transposing a vermilion triangle and cutaneous roll triangle from either side of the vertical scar (Fig. 21-5, B and C). If a stairstep deformity accompanies an underrotation of the medial lip, a major revision is preferred.

Vermilion

In all but the very minor forms of unilateral cleft lip, the vermilion below the cleft side of the bow is deficient in height. If this is not augmented with vermilion from the lateral lip element during the primary repair, a stairstep deformity of the red line (vermilion-mucosal junction) will be present, including central vermilion deficiency and exposure of mucosa (Fig. 21-6, A). The exposed mucosa is different in color from adjacent vermilion. Because it is not keratinized, it dries easily with resultant cracking, chapping, and peeling. Patients habitually lick the area or use lip balm. Although augmenting the vermilion as an isolated secondary procedure is difficult, it is usually possible as part of a major lip revision (see later section on Major Lip Revision). Augmenting deficient vermilion during a primary operation is easy; therefore primary prevention is preferable to secondary treatment. In a bilateral cleft lip, the vermilion of the prolabium is extremely short in height. If the central red lip is reconstructed using the red lip of the prolabium, as with a Manchester repair,⁸ or if the white roll flaps of the lateral lip elements were pared too medially

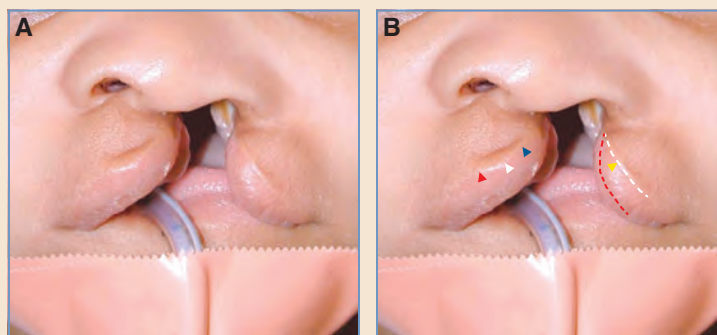


Fig. 21-6 A, A unilateral cleft lip and palate. B, The vermilion-cutaneous junction (*red dashed line*) and the cutaneous roll (*white dashed line*) of the lateral lip element are relatively parallel lateral to Noordhoff's point (*yellow arrowhead*). The two lines converge medial to Noordhoff's point. Cupid's bow on the noncleft side (*red arrowhead*), and the low point (*white arrowhead*) and planned position for the high point of Cupid's bow on the cleft side, are also shown.

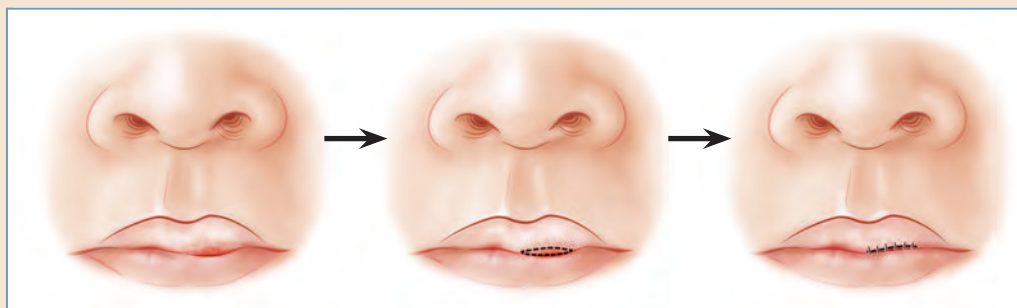


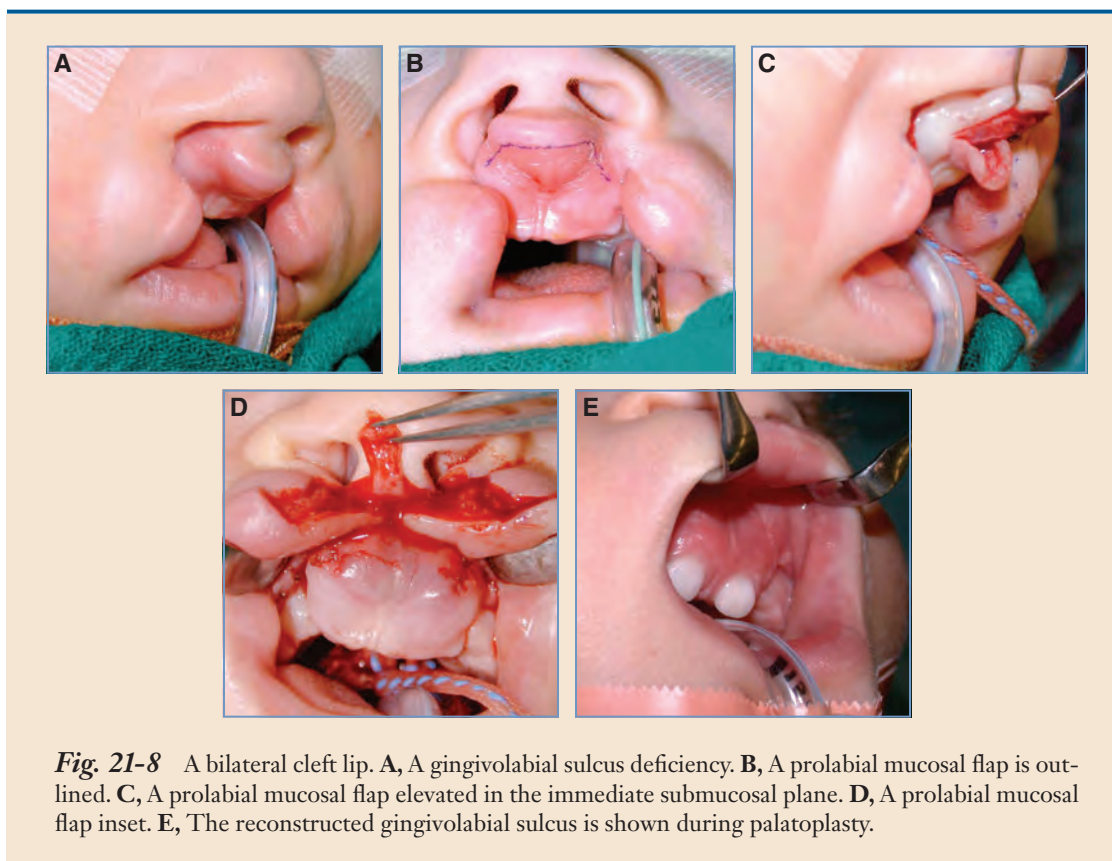
Fig. 21-7 Red lip revision by a transverse elliptical excision of mucosa preserves vermilion.

and carried with them vermilion of insufficient vertical height (see Fig. 21-5, C), central vermilion will be deficient and mucosa exposed. This situation generally requires a major lip revision.

The vermilion tends to heal nicely; however, the underlying scar can be excessive and add bulk to the free border of the lip. Some resolution of this swelling may occur long after the scar of the cutaneous lip has matured. If the swelling persists after 2 to 3 years, revision of the red lip should be considered. The red lip can be revised by either a transverse or vertical elliptical excision. A transverse reduction is often preferable (Fig. 21-7). Reduction of the excess by a transverse elliptical excision of mucosa behind the red line preserves vermilion and prevents a dog-ear deformity and extension of the scar onto the cutaneous lip.

Mucosa and Gingivolabial Sulcus

Primary surgery presents an ideal opportunity to create a generous and functional central upper gingivolabial sulcus⁹ (Fig. 21-8). A secondary deficiency of the central gingivolabial sulcus has both aesthetic and functional consequences; accordingly, numerous procedures for secondary correction of a gingivolabial sulcus deficiency have been described. Mucosal grafts may be required if no sulcus is present.¹⁰ For lesser deficiencies, an inverted-U-shaped flap of Erol and Ağaoğlu is effective.¹¹



Lateral Lip Deficiency

In the primary state, the transverse dimension of the cleft-side lateral lip (from the commissure to the peak of the Cupid's bow) is almost universally shorter than that of the noncleft side.¹² When the lateral lip is also deficient in height (from the subalare to the peak of the Cupid's bow), primary repair is more difficult. Because there is no way to increase the transverse length of the lateral lip, preservation of this length is required at the primary repair. Depending on the type of repair used, it may be necessary to further compromise the transverse length of the cleft-side lateral lip element to achieve the required vertical height. Inferior triangle repairs require less compromise of the transverse length than do rotation-advancement repairs, and they should be considered for repairing lips that have combined vertical and transverse deficiency of the cleft-side lateral lip.⁵

Volume

Augmentation cheiloplasty should be considered if the volume of an otherwise well-balanced lip is deficient. Whistle deformities generally are a combination of underrotation and central vermilion deficiency. These deformities are not corrected by volume augmentation. Many injectable filler preparations and implants are available, associated with various profiles of immune acceptance, retention, and duration of volume.² We prefer autologous free grafts to these preparations and favor the use of temporal fascia, as described by Chen et al.¹³ Temporoparietal fascia is preferable to dermal fat grafts, because it lacks dermal elements and the donor scar is inconspicuous.

Major Lip Revision

A major lip revision should be considered when the deformities described previously occur in combination. Takedown of the previous lip repair and complete revision of the repair with excision of all or most of the old scar can dramatically improve secondary deformities.^{14,15} When little tissue has been discarded and all the key landmarks are present, the previous repair can be treated much like a lip adhesion and excised within the lines of a definitive repair (Figs. 21-9 and 21-10). Occasionally, when this is not the case, an all-out effort should be made to preserve the existing key landmarks. If the key landmarks have already been discarded or distorted, an Abbé flap should be considered¹⁶ (Figs. 21-11 and 21-12).

The surgical goals of a major lip revision are similar to those of a primary repair²:

- Key landmarks are maintained.
- Tissue is thoughtfully discarded.
- Scars are placed along natural lines/seams of anatomic subunits.
- Muscles are aligned and repaired.
- The Cupid's bow is balanced.
- Symmetry is ensured.

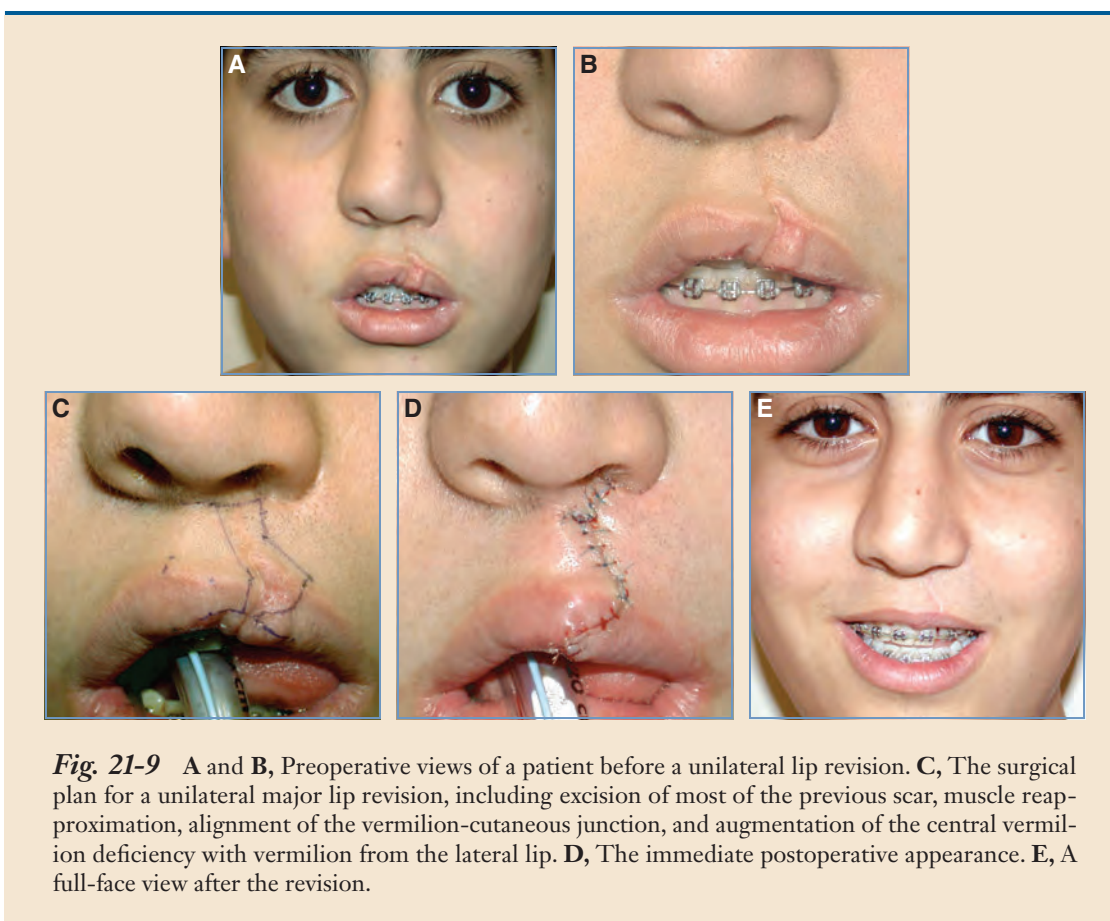


Fig. 21-9 A and B, Preoperative views of a patient before a unilateral lip revision. C, The surgical plan for a unilateral major lip revision, including excision of most of the previous scar, muscle reapproximation, alignment of the vermilion-cutaneous junction, and augmentation of the central vermilion deficiency with vermilion from the lateral lip. D, The immediate postoperative appearance. E, A full-face view after the revision.

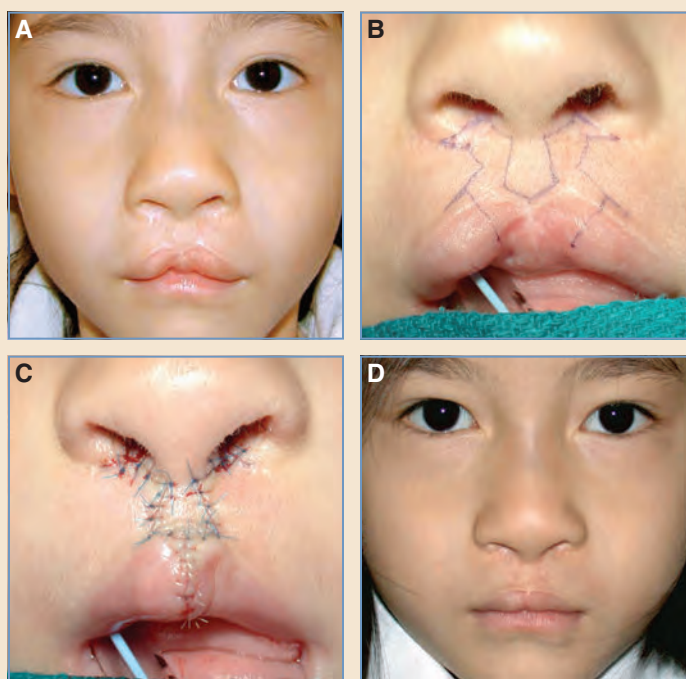


Fig. 21-10 A, A bilateral major lip revision. Preoperatively, the lateral lip elements have been paired medial to Noordhoff's point. A poor-quality cutaneous roll and a vermilion of inadequate height are retained. Mucosa is exposed below the central vermilion. B, The surgical plan includes excision of much of the previous scar; reduction of the prolabial skin to a small cutaneous shield; excision of the poor-quality Cupid's bow, exposed mucosa, and medial vermilion; the use of cutaneous roll- vermilion flaps designed lateral to Noordhoff's point; and muscle reapproximation. C, The immediate postoperative appearance. D, A full-face view 1 year postoperatively.

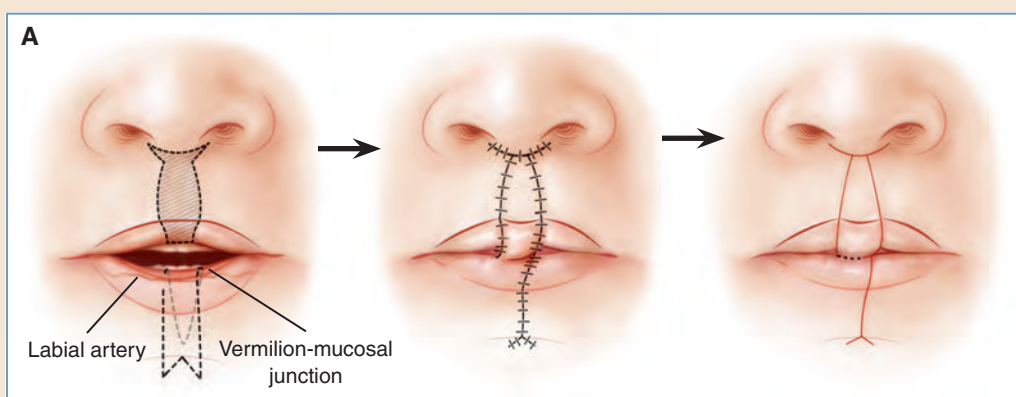


Fig. 21-11 A, An Abbé flap or "lip switch" allows replacement of deficient central lip, while reducing the hypertrophied lower lip. It also provides additional tissue for the columella and releases tension on the nasal tip.



Fig. 21-11, cont'd B-D, Preoperatively, the lip is short, tethered, and out of balance with the hypertrophied lower lip. The nose is tethered from the short columella. The overall impression is of significant class III skeletal relationships. E, The change in lip balance 1 week after division of the Abbé flap pedicle. The patient underwent a bilateral otoplasty simultaneously, and conchal cartilage was placed in the nasal tip in a configuration mimicking that of the normal alar cartilages. F and G, AP and lateral views show improved lip balance and, through soft tissue changes alone, a suggestion of corrected maxillary deficiency.

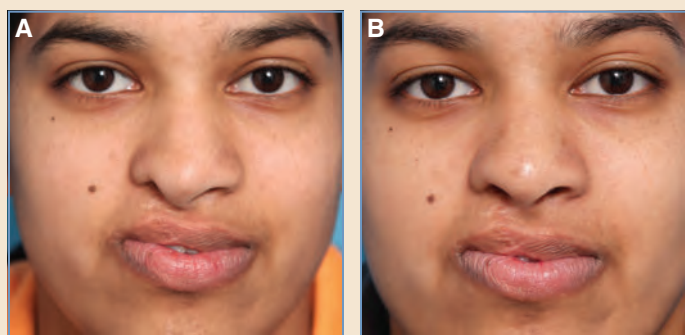


Fig. 21-12 Two cases are presented in which lip asymmetry and marked nasal asymmetry arose from early repairs and resulted in varying degrees of columellar displacement over the peak of the Cupid's bow. A, Although this patient's lip was well shaped, with well-repaired underlying muscle, it was short on the cleft side, and the nose was deviated to the cleft side. B, The lip was released from alar base to alar base along the nostril sill. This freed the columella to move toward midline and shifted the tissues to the right. The lip descended, matching the normal lip length and allowing correction without taking down the cutaneous scar.

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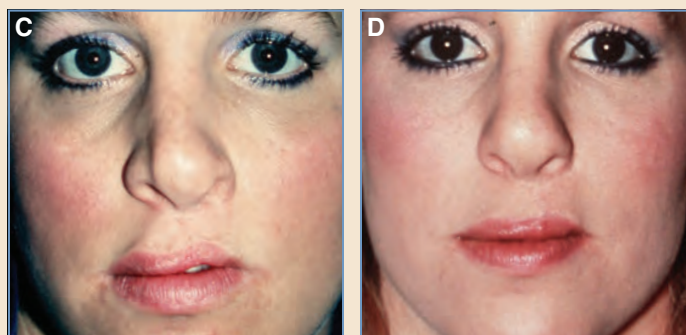


Fig. 21-12, cont'd **C**, This patient's columellar base was over the peak of the Cupid's bow on the noncleft side. The preoperative appearance suggested the nasal pyramid was markedly asymmetrical. The entire lip was taken down and revised using the basic principle of anatomic subunit repair. The lip shifted along the line from alar base to alar base, extra tissue from the nostril sill on the left moved across to the right, and the columella was allowed to move midline. **D**, The result of the revision suggests that a full rhinoplasty was performed. However, this atypical major revision of the lip with minor trimming of the cleft-side alar margin was all that was required after shifting the lip soft tissue and centralizing the columella.

SECONDARY CLEFT LIP NASAL DEFORMITIES

Secondary cleft nasal deformities vary significantly based on the extent to which each surgeon followed the lip repair principles described in Chapters 19 and 20 and addressed the nasal deformity during the initial cleft repair. Although some surgeons prefer to correct the nasal deformity when the underlying skeletal deficiency can be corrected, others think that adequate release of the nasal soft tissues, repositioning of the displaced cartilages, and filling of the deficient lining tissue helps to obtain excellent symmetry even in the presence of hypoplastic skeletal support.^{7,17-19} We have just seen how the correction of both minor and major secondary lip deformities can improve the residual nasal deformity. We will now focus on the anatomy of both unilateral and bilateral cleft lip nasal deformities, the steps by which the greater part of the deformity can be corrected early, the extent to which intermediate repair can further improve the deformity, and the maneuvers necessary to complete the reconstruction after skeletal maturity.

Primary Cleft Nasal Deformities

Unilateral Cleft Nasal Deformity

The anatomy of a *unilateral cleft nasal deformity* is the result of hypoplasia or distortion of the underlying bony and cartilaginous components and alteration of the soft tissue envelope, which affects the deformity to varying degrees^{7,17-21} (Figs. 21-13, *A* through *C*, and 21-14, *A*). The deformities include the following:

- The caudal septum deviates toward the noncleft side and is convex to the cleft-side airway.
- The cleft-side maxilla is deficient, and the cleft piriform margin is posteriorly positioned.
- Flaring of the lower lateral cartilage has, depending on the severity of the cleft, variably increased the angle between the medial and lateral crura.
- The cleft-side columella is foreshortened.

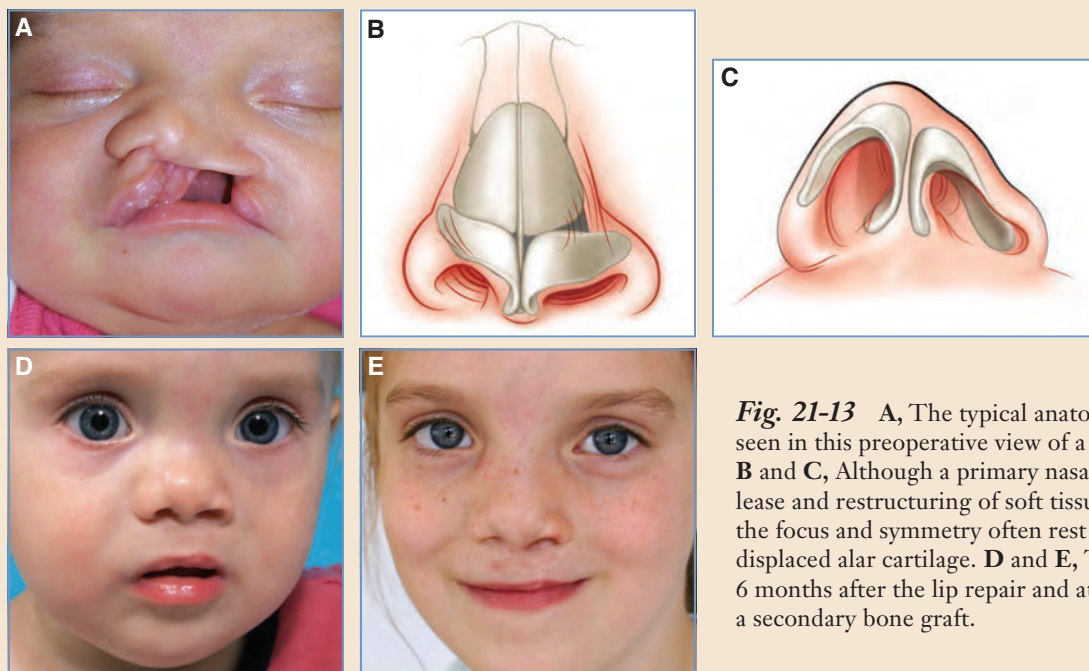
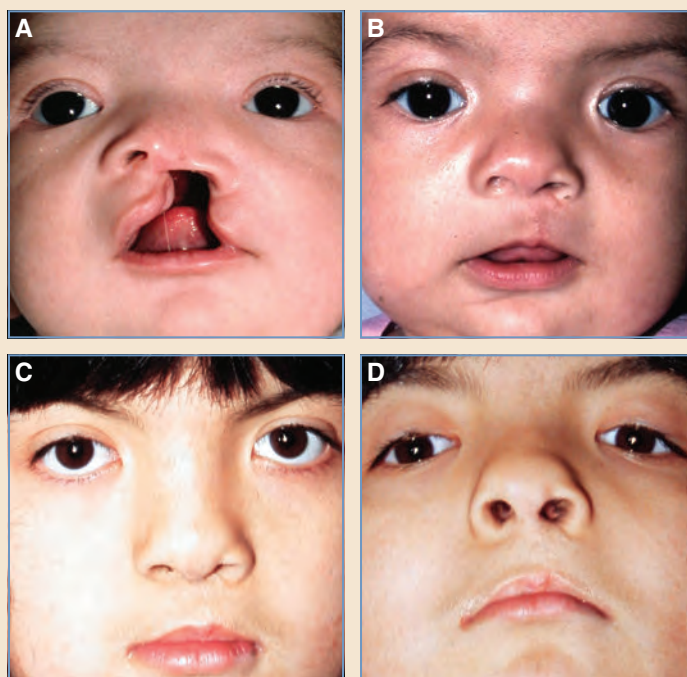


Fig. 21-13 A, The typical anatomic deformities are seen in this preoperative view of a wide unilateral cleft. B and C, Although a primary nasal repair involves release and restructuring of soft tissue and skeletal tissue, the focus and symmetry often rest on correction of the displaced alar cartilage. D and E, The patient is seen 6 months after the lip repair and at 6 years of age, before a secondary bone graft.

Fig. 21-14 A, This patient had a wide unilateral cleft lip and palate with a nasal deformity. B, Seven months after repair of the lip and primary nasal deformity, minor residual asymmetry of the nose persisted. C and D, By 6 years of age, no additional nasal repair has been performed, though she had a primary bone graft at 8 months of age. With growth the nasal symmetry has improved further.



The structural anomalies listed previously lead to the following external characteristics, which provide the stigmata of a unilateral cleft nose:

- The base of the columella deviates to the noncleft side.
- The cleft-side dome is retrodisplaced.
- The cleft-side nostril margin buckles inward.
- The cleft-side ala is flared laterally.
- The cleft-side alar base is posteriorly displaced.
- The interior cleft-side nostril is bowed by a vestibular web (linear contracture) from the apex to the piriform rim.
- The caudal septum rests within the non-cleft side nostril.

These characteristics can be corrected in a primary lip and nose repair to varying degrees depending on the severity of the deformity, the procedure chosen, and the skill of the surgeon. Our practice is to fully elevate the nasal skin off of the underlying cartilaginous structure and reposition it over the columella and ala. The lower lateral cartilage and its associated accessory cartilages are repositioned by dividing the anomalous connection of the lateral crus to the displaced piriform margin. The internal mucosal lining is lengthened with inset of the L-flap, allowing the alar base to descend into a more anatomically appropriate position. These steps improve alar symmetry to the noncleft side. Using quilting sutures along the alar crease, the nasal envelope and soft tissues are redraped in a way that reduces tension on the accessory cartilages and reduces the chance of relapse. These also improve the stereotypical bowing of the cleft-side ala and aid in the inset of the mucosal L-flap, if it has been used.

We think the most important part of the procedure is to ensure the soft tissues are released sufficiently to allow symmetrical placement of the alar base and to provide adequate lining to prevent later contracture. The practice of repositioning the caudal septum to help centralize the columella has been variously described.¹⁷⁻²¹ For some, it is a matter of routine to perform this as part of the primary repair of the lip and nose. It has been established that this does not restrict future growth of the nose or midface. It is not our standard practice in routine cases, but it may be beneficial in cases in which the width of the cleft or the deviation of the columellar base prevents tension-free approximation of the muscle (see Figs. 21-13 and 21-14).

Unilateral Cleft Nasal Deformity: Intermediate Repair

Primary rhinoplasty–intermediate repair is performed for residual cleft nasal deformity after the primary lip and nose repair and before the definitive rhinoplasty.^{18,22-24} Achievable goals include the following:

- The columellar base is centralized.
- The alar base is symmetrically positioned in the craniocaudal plane.
- The nostril margin is repaired to match the size and orientation of the non-cleft nostril.
- The lower lateral cartilage–accessory cartilage complex is further released from the lateral piriform margin.
- The vestibular web is released and repositioned using a right-angle Z-plasty (Fig. 21-15).
- Anteromedial advancement of the cleft-side lateral crus and dome
- A normal scroll area is created.

The corrections are limited, because the underlying skeletal deficiency is not corrected until the nasal base and piriform cleft are bone grafted. Once this is done, the definitive septorhinoplasty can be performed.

Occasionally, a primary repair was effectively performed, but either the L-flap was of inadequate size or secondary scar contracture along the nasal sidewall created a web that placed traction on the ala and flattened the nostril. This deformity can usually be addressed with a right-angle Z-plasty as early as the palate repair, possibly preventing an additional procedure when the child is of school age (see Fig. 21-15). The mainstay of our intermediate repair for unilateral cleft nasal deformity, however, is the repair described by Tajima and Maruyama,²³ which we modified and reported on in 1980.²⁴ The approach is directed at the nasal tip but can be combined with secondary repairs²⁴ (Fig. 21-16). Tajima and Maruyama's original description is modified primarily in the use of a cleft lip nasal retractor for placement of the supporting sutures to lift and reshape the displaced ala (Fig. 21-16).

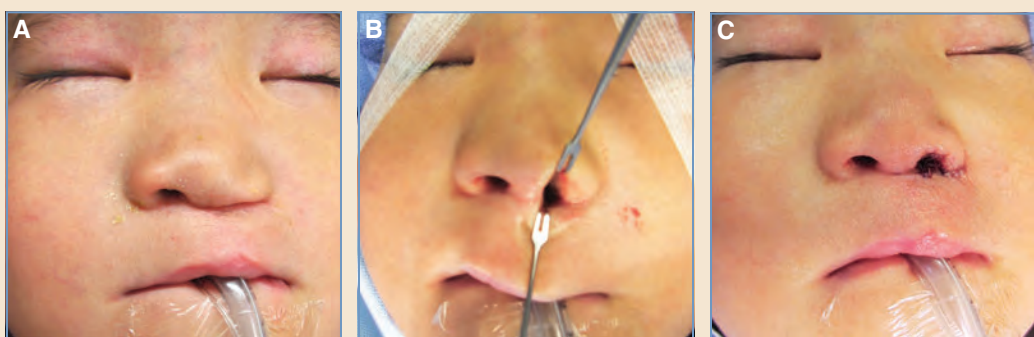


Fig. 21-15 A, The caudal displacement and flattening of the ala in this case appeared to be secondary to inadequate release and repositioning of the alar cartilage during the primary nasal repair. B, However, an inspection revealed a vertical alar web along the line of the inset L-flap. C, Release of the web with a right-angle Z-plasty repair allowed the ala to assume its normal position without additional suspension.

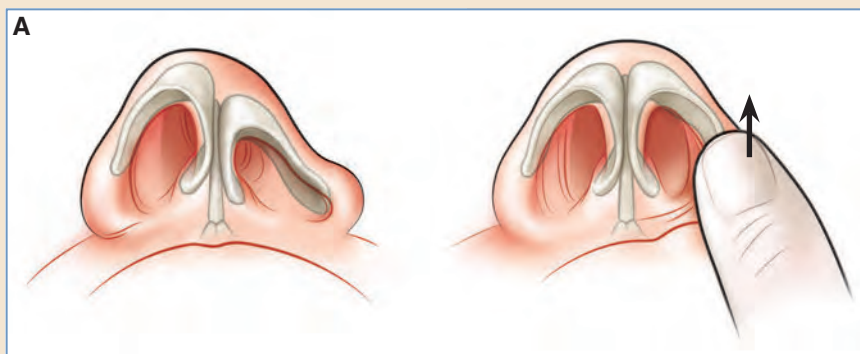


Fig. 21-16 The Tajima repair. A, With gentle pressure on the ala, the nostril is elevated and the position of the reverse-U incision is marked symmetrically with the non-cleft side.

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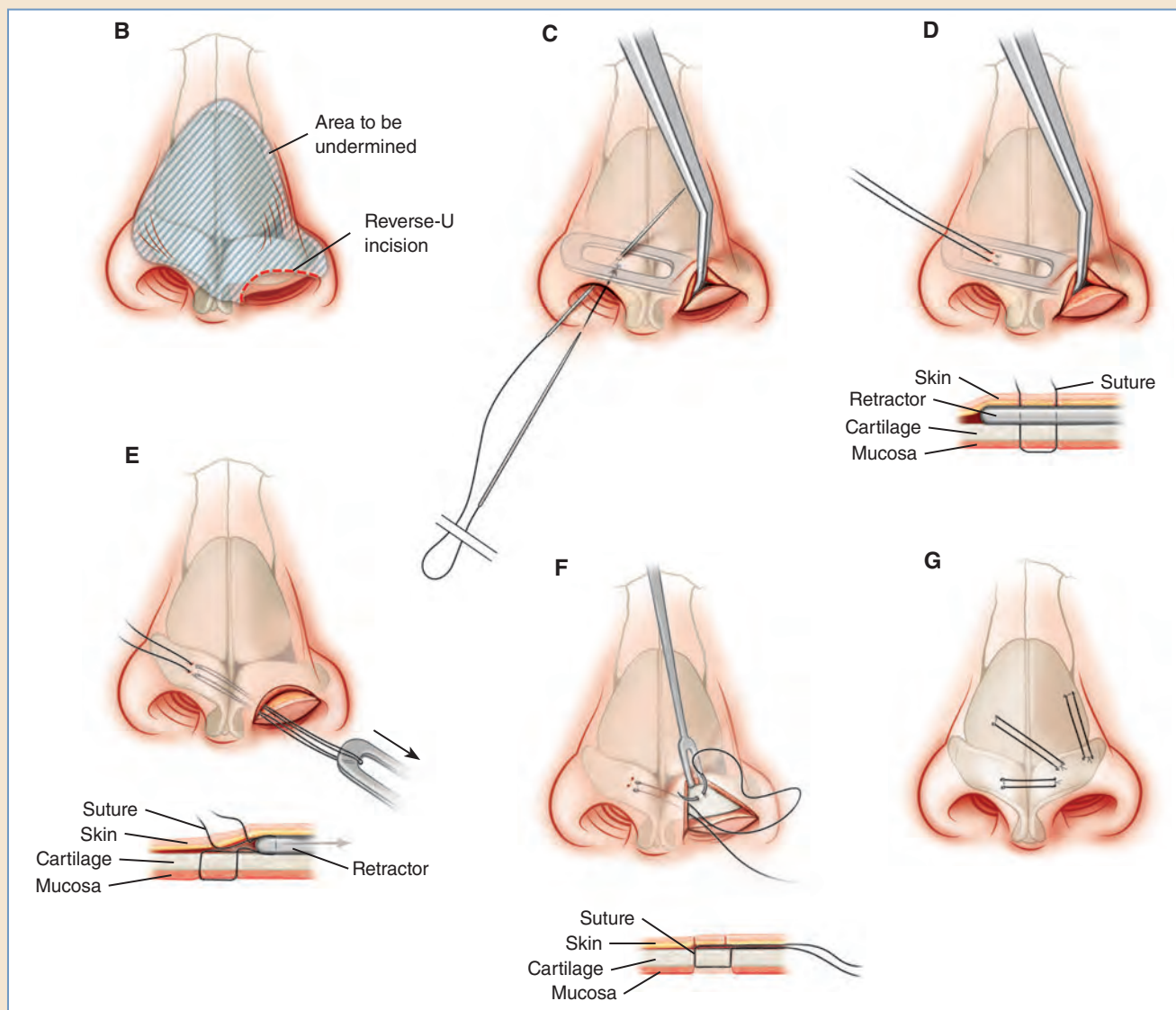


Fig. 21-16, cont'd B-F, A cleft nasal retractor is used for placing the suspending sutures, as shown in **K-O**. The skin is widely undermined, freeing the skin envelope from the underlying cartilage. The cleft nasal retractor is used to position the sutures for suspension of the displaced cartilage. The cleft retractor is placed between the cartilage and skin, and the sutures are passed with a straight needle through the mucosa, cartilage, retractor, and skin. The other end of the suture is passed in a similar fashion, starting 3 to 4 mm from the first one. The retractor is withdrawn and the free ends of the suture passed back through the skin and through the incision. One end is ready for placement through the cleft-side alar cartilage. **G,** The three suspending sutures used in a unilateral cleft nasal repair.



Fig. 21-16, cont'd H-J, The displaced cartilage is gently elevated, and the incision is outlined. The orientation of the incision is shown in three views. K, The skin envelope is freed extensively from over the upper and the lower lateral cartilages bilaterally. L-N, Clear nylon sutures are passed through upper lateral cartilage on the contralateral and ipsilateral sides and through the contralateral dome using the cleft retractor, as illustrated in B-F. O, Each of the three sutures is passed through the cleft alar cartilage: from the ipsilateral upper lateral cartilage to the lateral cleft ala, from the contralateral upper lateral cartilage to just lateral to the dome of the cleft ala, and from dome to dome. P, The sutures are tied, stopping each at the symmetrical position to the normal side. The extra tissue gained with the initial flap outline moves inward to provide the additional lining tissue. Q and R, The final correction.

Modified Tajima Reverse-U Repair

For minor deformities, we prefer to await facial skeletal growth and alveolar bone grafting for a definitive septorhinoplasty. Often, however, deformities of the soft tissues and the cartilaginous tip are significant, and patients can greatly benefit from earlier correction. Our timing relies on the severity of the deformity and personal social awareness of the patient. Typically, we perform primary repair using a modified Tajima approach^{23,24} (see Fig. 21-16) as early as 4 to 6 years of age.

An alar rim incision is planned by pushing gently on the cleft-side alar base to elevate the nostril. A reverse-U incision is planned at a location symmetrical with the non-cleft side nostril peak (see Fig. 21-16, *A*, and *H* through *J*). The incision starts at the membranous septum, curving slightly over the nostril rim, parallel to the cartilaginous dome, then reentering the nose, curving downward to end lateral to the fold in the nasal vestibule. The cartilage is identified and freed from the overlying skin, and a wide dissection is carried over the cartilaginous framework of the nose (see Fig. 21-16, *K*). Three 4-0 clear nylon sutures are passed through the bilateral upper and contralateral lower lateral cartilages and tagged (see Fig. 21-16, *B* through *F*, and *L* through *N*). This is facilitated by the use of a cleft nasal retractor inserted below the nasal envelope. The suture ends are passed through the mucosa, the cartilage, and the retractor (a modified Senn retractor with the tip drilled to allow passage of a Keith needle and suture) and then out the nasal skin using straight Keith needles. The suture ends are brought out of the alar rim incisions and then passed, using a curved, free needle, into sequential sections of the cleft-side lower lateral cartilage (see Fig. 21-16, *B* through *F*, and *O*). The sutures are then tied (see Fig. 21-16, *P*) simultaneously, ensuring proper tension to elevate the cartilaginous dome into an anatomic position (see Fig. 21-16, *Q* and *R*). Often, a slight excess of nostril skin is present on the mucocartilaginous cutaneous flap. This can be judiciously trimmed for a meticulous inset. The skin is closed with interrupted 6-0 chromic gut sutures and two or three quilting sutures along the alar crease.

Bilateral Cleft Nasal Deformity

Historically, bilateral nasal deformities were reconstructed in staged fashion using tissues recruited from the labial elements. In the early 1990s McComb²⁵ spearheaded an about-face movement to reconstruct the nose at the time of primary lip repair by bringing tissue from the nasal domes into the columella for bilateral defects. Since then, the technique has been refined and various presurgical orthopedic devices introduced to aid in positioning of the premaxilla, prolabium, and nasal domes.^{18,19,26,27} These devices are discussed in more detail in chapters addressing primary lip repair (see Chapters 19 and 20).

The anatomy of a bilateral cleft lip nasal deformity is not merely a duplication of the unilateral cleft deformity. The deformity has the following characteristics:

- The alar domes and medial crura of the lower lateral cartilages are splayed and caudally rotated.
- The superior columella is spread into the nasal tip by the laterally displaced and flattened medial crura.
- The lateral crura are positioned posteriorly along the deficient piriform margin.
- Excess fibrofatty tissue is interposed between the cartilaginous alar domes.
- The scroll area is missing, because the lower lateral cartilages are splayed laterally, away from a position overlapping the caudal upper lateral cartilages.

These structural anomalies lead to the following external characteristics of bilateral cleft nasal deformities:

- A broad, flat nasal tip
- A short columella
- An obtuse columellar-labial angle

- A blunted scroll area
- A nostril shape that is often slumped, with buckling of the alar margin and a widely splayed alar base

Repair of the Bilateral Cleft Nasal Deformity Using a Bilateral, Modified Tajima Reverse-U Technique in Primary Repair

The guiding principle in the primary repair of a bilateral cleft nasal deformity is to recognize that primary repositioning of the lower alar cartilages will reconstruct the nasal tip and columella. Our technique involves degloving the skin envelope of the nose during the primary lip repair and releasing the investing perichondrium of the lower lateral cartilages and their accessory cartilages from the insertion along the piriform margin. As in a unilateral cleft nasal repair, this allows re-draping of the skin once the cartilages are properly aligned. After repair of the lip, alar rim incisions are made at the prospective new apex of the nostril with a reverse-U pattern (Fig. 21-17, *A* through *E*). This varies from a unilateral Tajima technique in that the apex of the reverse-U shape should be more medially oriented to ensure a more natural tapering of the upper columella^{23,24} (Fig. 21-17, *E*). The nose is dissected until the lower lateral cartilages are identified and fully released from the overlying skin envelope. Often, the excess fibrofatty tissue needs to be thinned or removed to fully approximate the domes. Mattress sutures are placed at the medial crura and

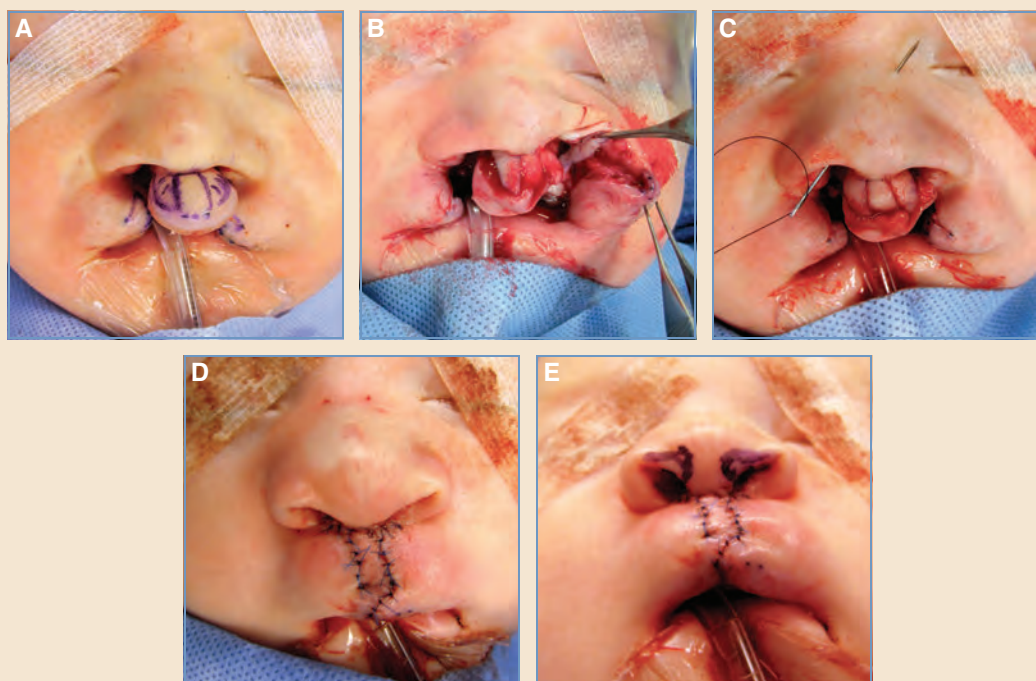


Fig. 21-17 A modified Tajima repair used for a primary bilateral cleft lip and nose. **A**, The outline of the lip incisions. **B** and **C**, An optimal repair requires adequate release of the lip and the displaced alar bases in the dissection and lining of the lateral nasal defects to ensure the final alar base positioning is not restricted. **D**, The lip repair is completed. At this stage, the columella is short, and the alar cartilages are still displaced. **E**, The reverse-U incisions are skewed more medially than in unilateral cases so that on completion of the repair, the columella tapers naturally.

Continued



Fig. 21-17, cont'd F-H, At the completion of the repair, the columella is lengthened. Note that the columellar-labial angle is well defined.

between the domes to bring the lower lateral cartilages into the proper position. Typically, this simultaneously redefines the scroll area. However, if the superiormost aspect of the lower lateral cartilages remains caudally displaced, mattress sutures are placed connecting the lower margin of the upper lateral cartilages to the lower lateral cartilages to elevate the tip appropriately. Once this is complete, there may be a small segment of skin on the reverse-U flap. This should be judiciously trimmed at the region of the soft triangles. Although this excision is often performed in bilateral Tajima repairs, it is rare in unilateral procedures. Overexcision can result in a thin and peaked alar margin that is very difficult to correct in future reconstructions. Quilting 6-0 chromic gut sutures are placed along the alar crease to set the skin envelope properly. Finally, a 5-0 chromic gut mattress suture is placed behind the columella to secure the mucosal flaps. The tissue gained with this approach results in a columella of normal proportion with a well-defined columellar-labial angle (Fig. 21-17, *F* through *H*).

Bilateral Cleft Nasal Deformity: Intermediate Repair

In cases in which the primary repair does not address the nasal deformity beyond the limited amount accomplished with the repair concentrated on the lip, the columella is short, the medial crura are placed under tension, the domes are flattened, and the tip is collapsed. This dramatically worsens the nasal deformity. A bilateral, Tajima reverse-U technique, previously described, can be used as an early nasal repair (Fig. 21-18). Early repair has several benefits. It improves the appearance of the nose and opens the external alar valves, which can be quite obstructed in this instance. The incisions lend themselves to the definitive open rhinoplasty. An early intermediate repair can also be used to better define the nasolabial angle. Finally, a chonchal tip graft can help to further refine the tip if intradomal mattress sutures fail to do so (Fig. 21-19). At the definitive repair, this extra cartilage often aids the surgeon by providing added cartilaginous tissue to support tip sutures or to smooth strut grafts. The main differences between a unilateral and a bilateral modified Tajima repair used as an intermediate repair and a repair carried out primarily is the orientation of the reverse-U incision (angled medially in bilateral cases) and the number of sutures placed (see Figs. 21-16 *A*, *I*, and *J*, 21-17, *E*, and 21-19, *B*).

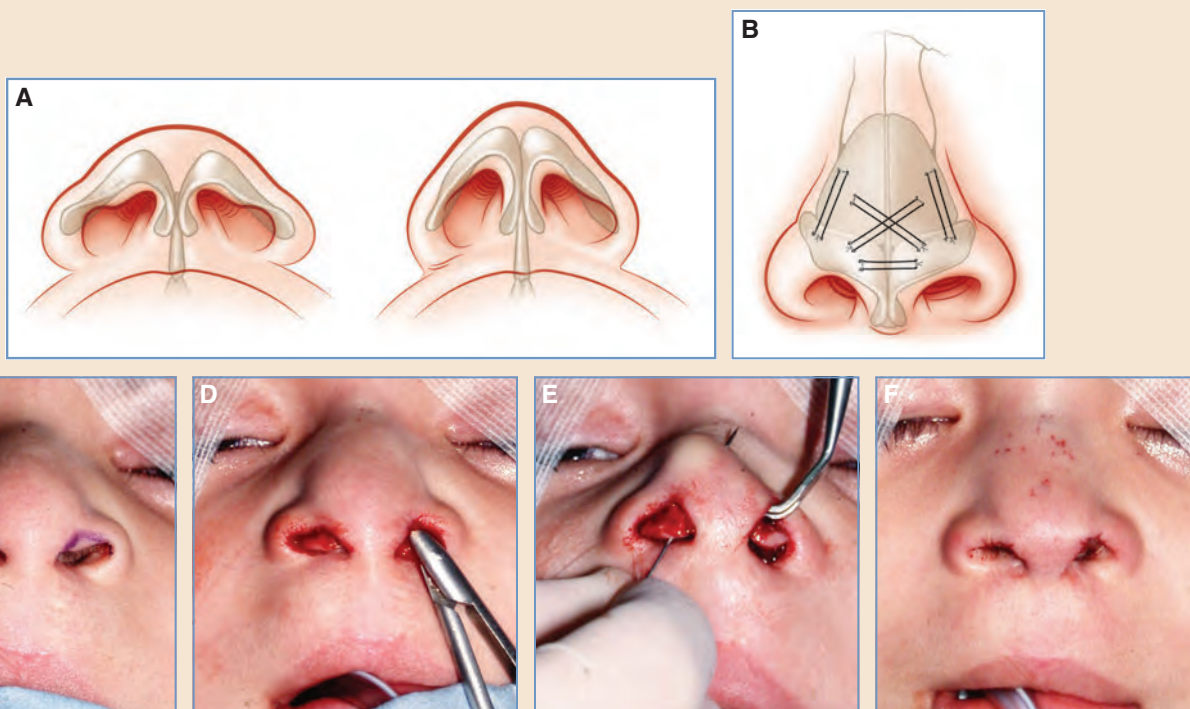


Fig. 21-18 The application of the bilateral Tajima repair in early secondary correction. **A**, The difference in orientation of the alar cartilages between an uncorrected bilateral cleft nose and a normal nose. **B**, The position of the suspending sutures. **C**, The incisions in this case are more typical of those used in a unilateral case; that is, they are not skewed medially. **D**, The wide undermining of the soft tissue envelope off the cartilages. **E**, Suspend sutures are passed using a cleft nasal retractor. **F**, The reshaped tip after tightening of the sutures and closure.



Fig. 21-19 In selected cases, the nasal tip shape and projection can be augmented with a conchal cartilage graft placed just above the domes of the alar cartilages as they are approximated. **A**, A preoperative view shows the broad, flat nasal tip. **B**, Incisions taper superiorly, as described previously, and conchal cartilage is shaped for placement. **C**, The completed repair.

The supporting sutures pass from the lateral lower lateral cartilage to the ipsilateral upper lateral cartilage, from the lower lateral cartilage just lateral to the dome to the contralateral upper lateral cartilage, from dome to dome, and from medial crura to medial crura. The passage of these sutures is facilitated with a cleft nasal retractor. As the sutures are tied, the nostril changes orientation from horizontal to oblique, the columella tapers, and the nasolabial angle sharpens. This change of orientation helps to gain the additional columellar length, from the nose rather than the lip, which results in the need for minor trimming of the skin of the reverse-U flaps.

Secondary Cleft Nasal Deformities

The goals of secondary cleft rhinoplasty are to create a symmetrical, normal- and natural-looking nose from the top down and to optimize nasal breathing and function. Although primary cleft rhinoplasty can do much to achieve a more normal-looking nose, the limitations in dissection and bony and septal work can lead to secondary deformity as a patient enters the teenage years.

Description of Secondary Cleft Nasal Deformities

Secondary cleft deformities occur in all shapes and sizes, but the general deformity is fairly uniform and includes the following:

- Skeletal deficiency of the alar base
- Nasal bone pyramid asymmetry and excess width
- Nasal tip asymmetry, with shortening, flattening, and malposition of the affected side lower medial and lateral cartilages (see Fig. 21- 18, *B*, and *D* through *F*)
- Alar base soft tissue asymmetry/deformity
- A thick skin/soft tissue envelope
- Septal deviation

Myriad associated iatrogenic deformities can be present depending on the type of primary repair performed.^{2,21-25}

Septorhinoplasty in the Cleft Patient

The timing of secondary rhinoplasty and septoplasty in cleft nasal patients varies, but these procedures are typically performed when patients are teenagers. We do not rely on a single standard age for repair, but instead usually rely on the following guidelines^{28,29}:

1. The patient is at least 1 year postmenarcheal.
2. Growth has not been significant over the past 1 to 2 years, as documented by the patient's pediatrician.
3. The patient's parents are ready for the patient to proceed with surgery.

These guidelines have been shown to produce consistent, lasting results, establishing that there is no single, set age at which rhinoplasty can be safely performed. We will perform septorhinoplasty even earlier, in the preteen and early teen years, if the deformity is severe enough to cause significant psychological detriment.

During cleft nasal and septal surgery, surgeons may encounter both natural and iatrogenic deformities. Every nose has a slightly different (if not majorly different) morphology. Therefore, for all deformities, surgeons should focus on deconstructing the nasal tip and doing whatever is required to make it look normal and then complete the dorsal and bony work (Fig. 21-20). This is accomplished through soft tissue release, repositioning of the native structures, suture techniques, and cartilage grafting. The same principle applies to the septum. The tip or septum should be completely replaced and rebuilt if the deformities are too severe to salvage.

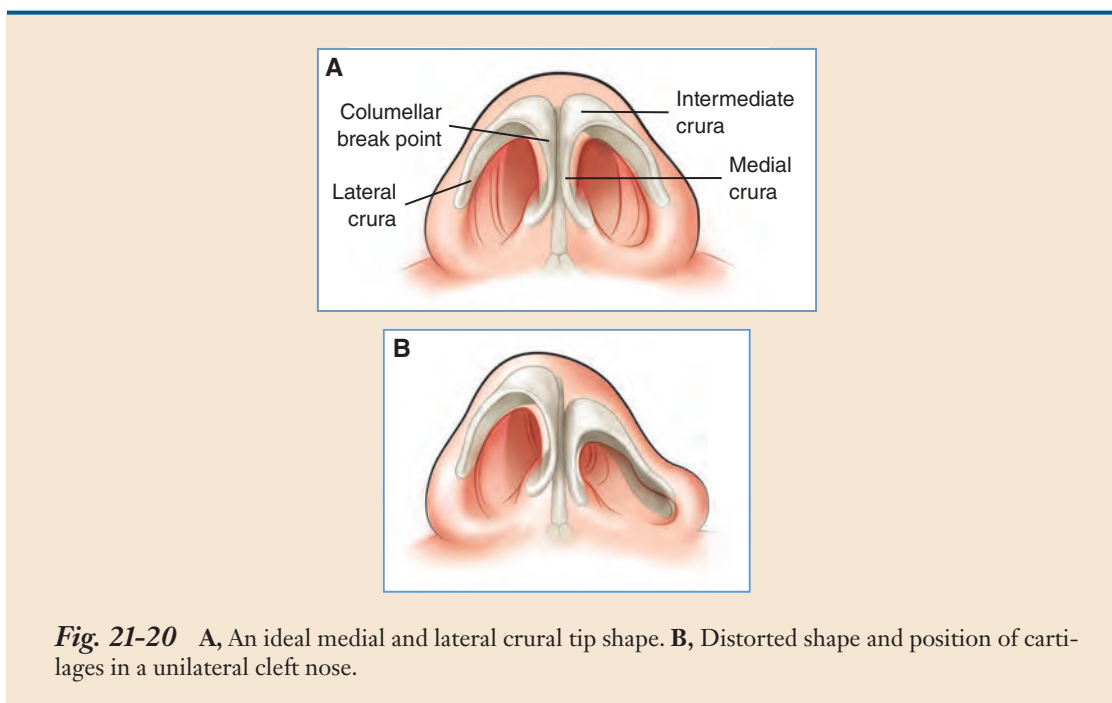


Fig. 21-20 A, An ideal medial and lateral crural tip shape. B, Distorted shape and position of cartilages in a unilateral cleft nose.

Surgical Technique

Details for each of the operative steps are listed below, but the major overview concepts include septoplasty/septal reconstruction, alar base augmentation, reconstruction of the nasal tip cartilages and lengthening of the columella, evening and narrowing of the nasal bones, creating the ideal dorsal profile, and creating a more symmetrical alar base, combined with any necessary alar soft tissue excision techniques. The sequence of these steps may be debated, but the most consistent and logical order is as follows:

1. The nose is opened, and the nasal tip is deconstructed.
2. Septoplasty/septal reconstruction is performed.
3. The columella is lengthened, and the tip is reconstructed and stabilized.
4. The dorsal profile is refined to match the tip position.
5. Osteotomies are completed.
6. The alar base and tip soft tissue are refined.

Septoplasty

Septoplasty is performed through an open approach in cleft patients. The septal mucosal flaps can be easily elevated starting at the caudal end of the septum, between the medial crural cartilages of the tip (Fig. 21-21). Once the upper lateral cartilages are separated from the septum, the entire septum can be visualized using a nasal speculum. This is especially useful with severe or complicated deviations that require careful elevation of the mucosal flaps to prevent rents and tears.

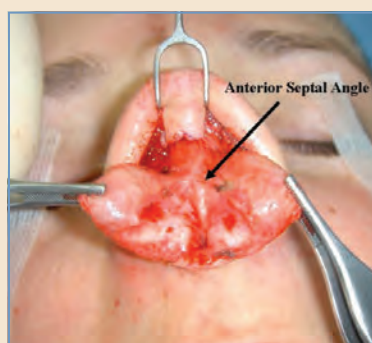


Fig. 21-21 The septum is often most easily accessible after the anterior septal angle is identified.

With the entire septum visualized, a portion of the posterior quadrangular cartilage can be excised. (A No. 15 blade is much preferred over a swivel knife.) All posterior bony septal deviations can be directly excised. A dorsal and caudal portion of the septum will remain, which should ideally be at least 1 cm wide (or more if a dorsal hump takedown is planned), thus creating a stable L-strut.

Remaining septal deflections, whether caudal or dorsal, may require further steps to ensure they are completely straight. Curvatures of the dorsal septum create asymmetry of the brow-tip aesthetic lines on an AP view, a severely canted dorsal septum can lead to a persistently crooked nose, and residual caudal deflections can lead to airway obstruction. Options for correction include the following³⁰:

1. Scoring on the concave side to curve the cartilage away from the scored side
2. Castellation of a dorsal strut from above and below (vertically incising the septum yet keeping it intact)
3. The placement of upper lateral cartilage-to-septal transfixion sutures
4. The placement of a cartilaginous spreader graft on the concave side of the septum
5. The placement of dorsal onlay graft to camouflage the curve
6. Total or subtotal resection of the deformed septum and reconstruction with a septal or rib cartilage graft or grafts

We tend to rely heavily on subtotal and total reconstruction of the deformed septum to ensure a straight and consistent result, which has reduced the revision rate. Remaining caudal septal deflections require a swinging-door approach, which entails completely releasing the caudal septal base from the underlying maxilla, excising a 1 mm portion if necessary, repositioning the base over the direct midline maxillary crest, and fixing it in place with mattress sutures.

Tip Reconstruction

No matter what deformities are encountered during cleft rhinoplasty surgery, the goal should be to do whatever it takes to make the tip structures look normal.⁴ We start by reconstructing the tip to achieve three ideal, major nasal parameters: length, projection, and rotation of the tip.^{22,24,31-35} A superbly harmonious result requires ensuring the major nasal parameters are achieved and set in the desired position before addressing the remaining nasal issues. The dorsum can then be tailored to fit these parameters and the fine tip work details completed. We have found that approaching the nose in this manner will guide the surgeon to the most predictably acceptable outcome.³⁶

Using an open approach, we make an inverted-V incision with vertical incisions along either side of the columella. Unlike the standard approach in a noncleft rhinoplasty, the lateral columella incision is extended inferiorly along the anterior edge of the medial crural footplate on the affected side and carried down into the nasal sill (Fig. 21-22). With the nasal flap elevated and the tip cartilages exposed, the supporting ligaments of the tip are released to allow full mobilization of the tip structures. It is easiest to gain access to the septum at this point, and the septoplasty is performed. Once the septoplasty is completed, the medial crural footplate on the affected side is completely freed and advanced upward to meet the ideal position of the dome on the nonaffected side. This rotation-advancement flap of cartilage and mucosa is critical to ensure tip symmetry and a consistent, long-lasting result.

With the columellar base reasonably exposed, the alar base can be augmented. We typically use harvested septal or rib cartilage³⁷⁻⁴¹ and place the graft through a precise pocket exposed through the access provided once the medial footplate has been released. The broad, amorphous tip is then addressed. Cephalic trim and evening of the lower lateral cartilages are performed. A double hook is used (with one tong under each dome and using the instrument to stabilize the tip position) to set the ideal length, projection, and rotation. A columellar strut is placed to stabilize the ideal nasal tip parameters and to set the dome position securely (Fig. 21-23). The ideal dome position is then marked. Intradomal (single-dome-unit binding) sutures are placed using 5-0 or 6-0 permanent suture.

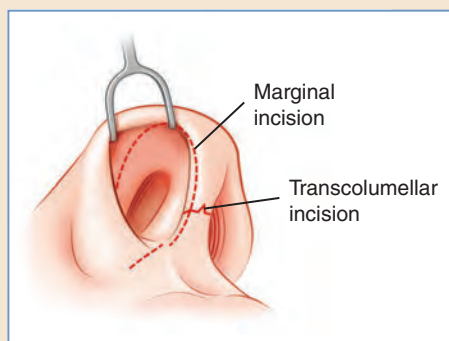


Fig. 21-22 Unlike the standard approach in a noncleft rhinoplasty, the lateral columella incision is extended inferiorly along the anterior edge of the medial crural footplate on the affected side and carried down into the nasal sill.



Fig. 21-23 A double hook is used to set the ideal length, projection, and rotation. (One tong is placed under each dome, and the instrument is used to stabilize the tip position.) A columellar strut is placed to stabilize the ideal nasal tip parameters and to set the dome position securely.

With the columellar base and tip stabilized, the surgeon can examine and reconstruct the lower lateral cartilages for symmetry and shape. This almost always requires bilateral lateral crural strut grafts to be placed. Lateral crural strut grafts will even the lower lateral cartilages and reduce the convexity (bulbosity), thereby creating an ideal normal shape. The strut grafts will provide long-term stability to the repair. The mucosa is dissected free from the undersurface of the lower lateral cartilages. On the nonaffected side, the strut graft is placed beneath the native lower lateral cartilage and secured. With a unilateral cleft deformity, the lower lateral cartilage attachment laterally to the soft tissues is asymmetric. Therefore, on the affected side, the lateral lower lateral cartilage is completely released from its soft tissue attachments, an extended alar strut graft is placed, and the lateral end of the extended alar strut graft is placed into a new, more caudal position by dissecting a new pocket and fixating the graft over the piriform aperture. This may also be required on the nonaffected side to even the alar rim positions bilaterally. The entire tip complex looks normal and symmetrical, and tip cartilage grafting can be performed to achieve the ideal projection and tip shape at the domes. Not every patient requires tip grafting. It should be done only when necessary to prevent overprojection or an abnormal, grafted-tip appearance. The length, projection, and rotation have been set. The dorsal profile can be set to the ideally positioned tip.

Correction of a short, overly rotated nose in a bilateral cleft patient typically requires rib cartilage grafting and nasal lengthening.⁴² The nose can be lengthened by infratip lobule grafting or by augmenting the nasofrontal region. More severe cases may require bilateral extension spreader grafts with a columellar strut graft or caudal septal extension grafts. Dorsal augmentation in cases of saddlenose deformity produces apparent lengthening and will result in a more aesthetically pleasing, straight dorsal profile.

Dorsum Techniques

At this point, the tip should be symmetrical with a normal appearance. It is time to match the dorsal profile and appearance to the tip. The nasal dorsum requires a tremendous amount of judgment in rhinoplasty. The nasal dorsum comprises the middle vault (cartilaginous) and the upper nasal vault (osseous). Pollybeak deformities are either cartilaginous or secondary to soft tissue excess and should be corrected to achieve a straight dorsal profile. The osseous vault requires osteotomies. Additional deformities anywhere along the dorsal length (that is, open-roof deformities and upper lateral cartilage contour deformities) require correction. Bilateral cleft deformities may require central onlay dorsal augmentation grafting. As stated previously, no matter the deformity, the goal should be to undertake whatever maneuvers are necessary to straighten the dorsum.

In patients with dorsal fullness, creating a straight dorsal profile requires leveling the bone, the bony/cartilaginous junction (rhinion), the dorsal cartilaginous septum (pollybeak), or all three. The bone is rasped, starting with a more coarse rasp, followed by finer rasps. The cartilaginous pollybeak is corrected by leveling the dorsal quadrangular cartilage to a favorable profile using sharp straight scissors or a No. 11 blade. The dorsum is augmented with cartilage grafting, either using septal cartilage (single or stacked) for smaller concavity deformities or rib cartilage for more severe deformities requiring a thicker graft. Spreader grafts set above the profile of the dorsal cartilage can be used to augment the dorsum through the middle third.

Nasal bone anatomy is quite variable. Some patients have short nasal bones; others have long nasal bones. Younger patients tend to have very thick nasal bones. Regardless of the nasal bone

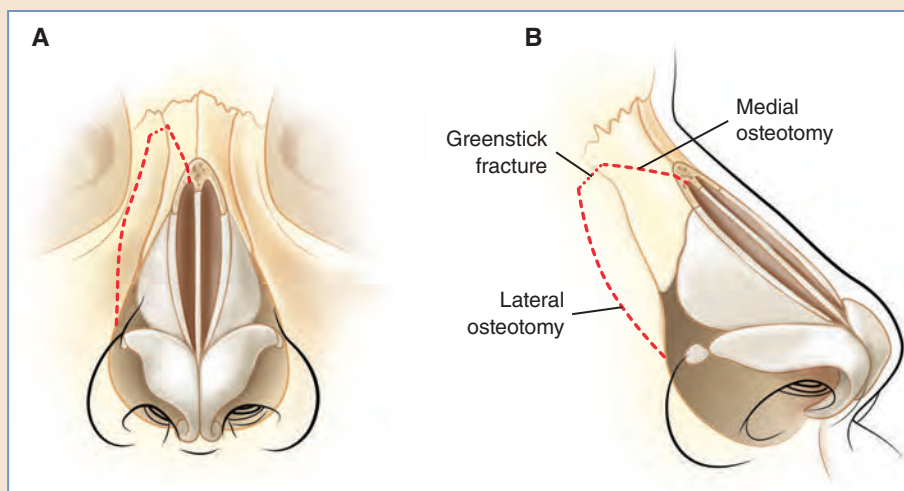


Fig. 21-24 Nasal bone osteotomies. **A**, Medial osteotomies are performed first, with care not to disrupt the septal–nasal bone junction. **B**, Lateral osteotomies are started superior to the inferior turbinate and performed in a high-low-high fashion. An osteotome can be used as a lever to assist in making a greenstick fracture of the nasal bone medially.

anatomy, medial and lateral osteotomies are required to narrow the upper third and straighten the nose.^{43,44} We use guarded straight and curved osteotomes. A medial osteotomy is performed first, starting at the caudal end of the nasal bone, between the septum and upper lateral cartilage on each side. The osteotomy should be faded and curved laterally as it is continued cephalad, preventing a rocker deformity. Lateral osteotomies are then performed, using an internal approach, with the osteotomy starting just above the head of the inferior turbinate, along the lateral nasal wall. A high-low-high osteotomy is thus effected. The medial and lateral osteotomies are not joined, but rather the lateral osteotomy is brought just medial to the medial canthus, and a greenstick fracture is produced using the force of the osteotome to create an infracture. A greenstick fracture will result in a much more stable fracture site without excessive bony instability (Fig. 21-24). Spreader/spacer grafting is then performed as needed to ensure straightness of the nose on an AP view and/or to correct internal nasal valve dysfunction. The upper lateral cartilages are finally secured back to the midline using an absorbable suture.

The overall underlying structures should be straight and ideally positioned. Concavities or irregular depressions can be evident anywhere along the dorsum and/or tip. Often, dorsal irregularities need to be camouflaged with onlay grafting using cartilage, soft tissue, or fascia. Cartilage grafts should be thin and beveled as much as possible so that they do not show through thin skin once the edema resolves and to prevent secondary step-off deformities. These grafts can be secured directly to the underlying skeleton framework, or they can be secured more precisely using transcutaneous sutures, which can be left in place for 4 to 7 days without external scarring.

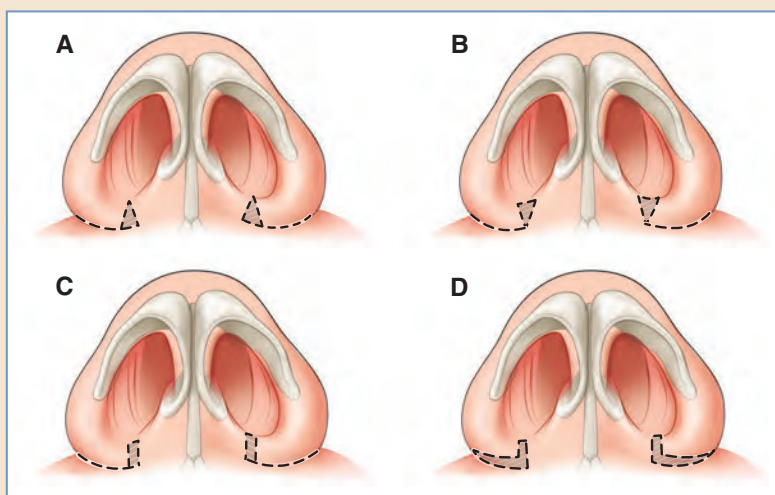


Fig. 21-25 **A**, Sill excision for external base width reduction only. An inverted-V excision is used to decrease the alar base width without changing the nostril circumference. **B**, Sill excision for nostril diameter change only. A V excision is made to decrease the nostril circumference without changing the base width distance. **C**, Sill excision for combined alar base width reduction and nostril diameter reduction. **D**, Alar flare reduction. A portion of the lateral alar base is excised to decrease the tip-alar facial groove distance.

Alar Base Soft Tissue Techniques

Once the overall major parameters of the tip, the fine structural tip detail, and the nasal dorsum have been optimized, the nasal tip soft tissues are evaluated and treated. The following features are evaluated and treated (Fig. 21-25):

- The alar base width
- Nostril diameter and symmetry
- The alar flare
- Alar hooding
- Alar retraction
- A thickened skin-soft tissue envelope

Alar Base Reduction

Understanding that the ala has a cutaneous surface and a vestibular surface is essential. This helps to maintain or narrow the width of the alar base and to maintain or narrow the nostril size separately in well-controlled fashion. We prefer to use the terms *external alar base reduction* and *internal alar base reduction* to distinguish between the two different surface reductions (alar base width and nostril diameter, respectively). An external alar base reduction is performed to reduce the columellar-alar base distance, thereby reducing the apparent width of the lower third of the nose on AP and basal views. An internal alar base reduction is performed to narrow the nostril circumference (vestibular surface).

An alar base reduction is best performed in the nasal sill. Not disrupting the natural curve of the alar base is critical. If only an external reduction is planned (to reduce the columellar-alar width), the appropriate estimation of resection is planned and marked along the upper and lower portion of the nasal sill, and the excision is performed. The excision is wide at the bottom of the sill and tapered in an inverted-V pattern to address only the external reduction without changing the circumference of the nostril. If the nostril circumference needs to be reduced, the excision should not be tapered on the internal edge of the sill, and the appropriate amount of tissue should

be excised.⁴⁵ In most cases of alar base reduction, it is necessary to create a true advancement-rotation flap of the entire alar base to prevent notching, to provide a more natural curve, and to reduce tension along the suture line, thereby optimizing the scar outcome.

Alar Flare Reduction

With a rotation-advancement flap closed with a single suture, alar flaring can be assessed. If necessary, an alar flare reduction is performed. Alar flaring is seen when the lateral aspect of the ala extends significantly beyond the alar-facial groove. If alar flare reduction is indicated, the amount of resection is planned, marked, and excised. The wounds are completely closed with 6-0 nylon. Most of the cutaneous sutures are removed on postoperative day 4, with the exception of the central sill suture and the more cephalad sutures on the inner portion of the sill, which are left in place for 6 days. Steri-Strips are placed for an additional 4 days once the sutures are removed.

Alar Hooding Reduction

Alar hooding is best analyzed from a direct lateral view. It is seen when the most caudal curve of the ala (alar rim) hangs excessively low, either from excessive bulk of the ala or from an excessively caudal alar insertion, thus hooding over the columella and obscuring it from view. If indicated, the resection is planned using a direct lateral view. The visual border of the ideal alar curve is marked, and an ellipse is drawn on the inner portion of the ala that matches the outer visual border. This incision should be planned carefully and should curve gently to prevent distortion of the nasal ala. After the excision, the wound is closed with running 6-0 nylon along the most caudal aspect of the alae. The suture line falls along the visual border, in a natural plane. We have not found healing along this line to be a problem. Sutures are removed on postoperative day 4.

Alar Base Repositioning

If the vertical height of the lip is in balance (that is, if the Cupid's bow is level and the subalar-Cupid's bow peak heights are symmetrical), minor asymmetries of the alar bases in the vertical dimension can be addressed by a Z-plasty. If the alar insertion is too low, reciprocal transposition of the alar base with a triangle taken from cephalad within the nostril sill and vestibule can be helpful; however, care must be taken not to introduce skin of different character (thickness, color, hair, or scar) onto the upper lip (see Fig. 21-2). If the alar insertion is too high, a reciprocal transposition of the alar base with a triangle taken from the upper lip can be performed.

Thickened Skin-Soft Tissue Envelope

Once a nasal flap has been elevated in an open approach, the soft tissue under the flap can be conservatively thinned using sharp scissors. Only the tissue that is excessively hanging should be excised. Excessive thinning of the undersurface of the dermis can lead to ischemia of the flap.

FUNCTIONAL NASAL ISSUES AND TURBINATE CORRECTION

The structural components of the airway comprise the septum, internal nasal valves, external nasal valves, and inferior turbinates, all of which require examination and optimization and can be corrected surgically.⁴⁶ Repair of the nasal septum through septoplasty and spreader graft placement to correct internal nasal valve dysfunction has already been described in this chapter. The external nasal valves will be strengthened and optimized when lateral crural strut grafting is performed to even the tip complex. After these procedures and a rhinoplasty have been performed, enlarged inferior turbinates can be reduced. It is best to reduce the overall size of an inferior turbinate with mucosal reduction outfracture of the bone. Complete excision should be avoided. Our preferred technique is submucous resection of the mucosa using a turbinate debrider with a direct outfracture. Coblation and cautery can also be used (Figs. 21-26 through 21-28).

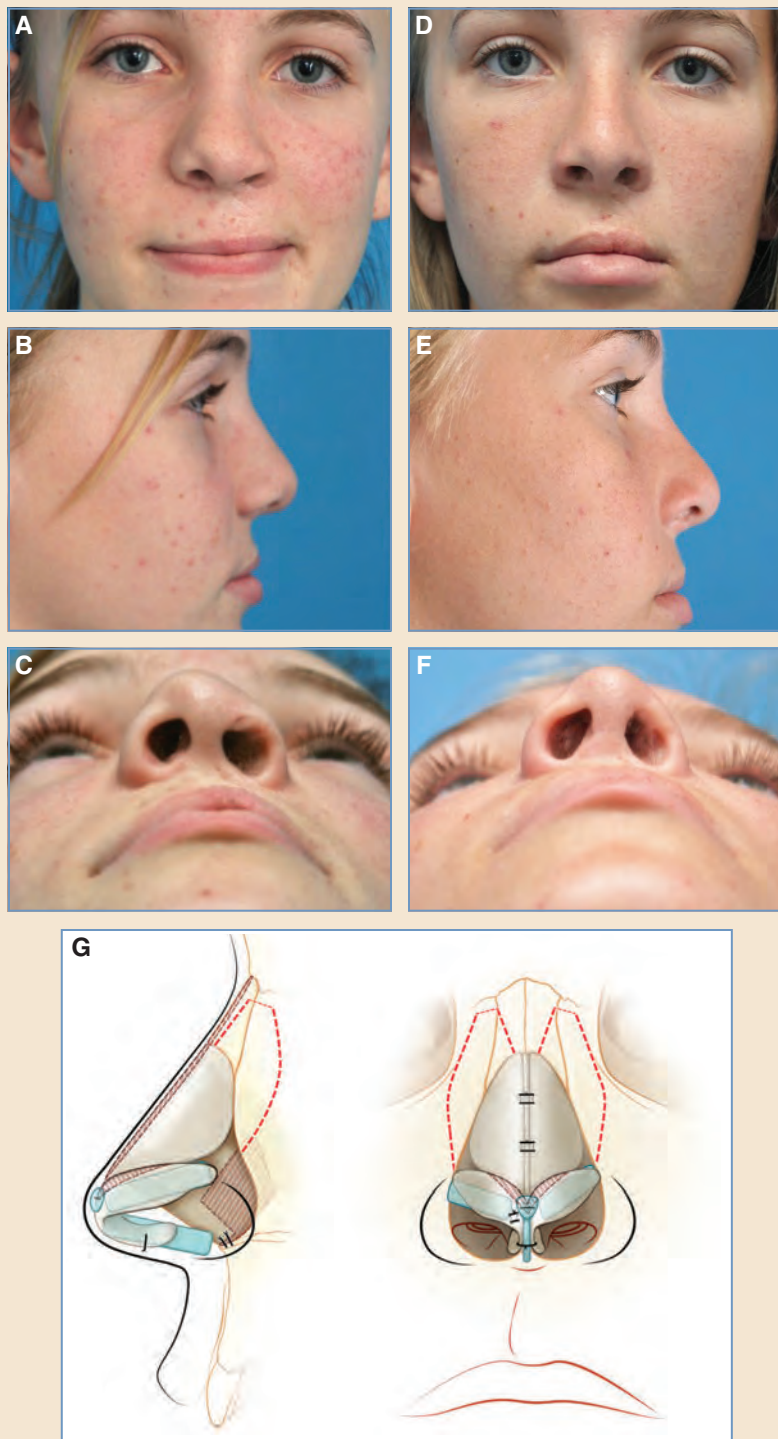


Fig. 21-26 This unilateral cleft patient underwent septorhinoplasty using an open approach with a unilateral extended columellar incision, as described previously. **A-C**, Preoperative views. **D-F**, One-year postoperative views. **G**, Rib graft was harvested, given the paucity of septal cartilage after the previous septoplasty. The caudal septum was repositioned. The tip was reconstructed with bilateral lateral crural strut grafts (the right lateral crus with a strut graft was repositioned inferiorly), a columellar strut, a tip graft, interdomal and intradomal sutures, and cephalic trim. The dorsum was rasped. Bilateral medial and lateral osteotomies were performed.

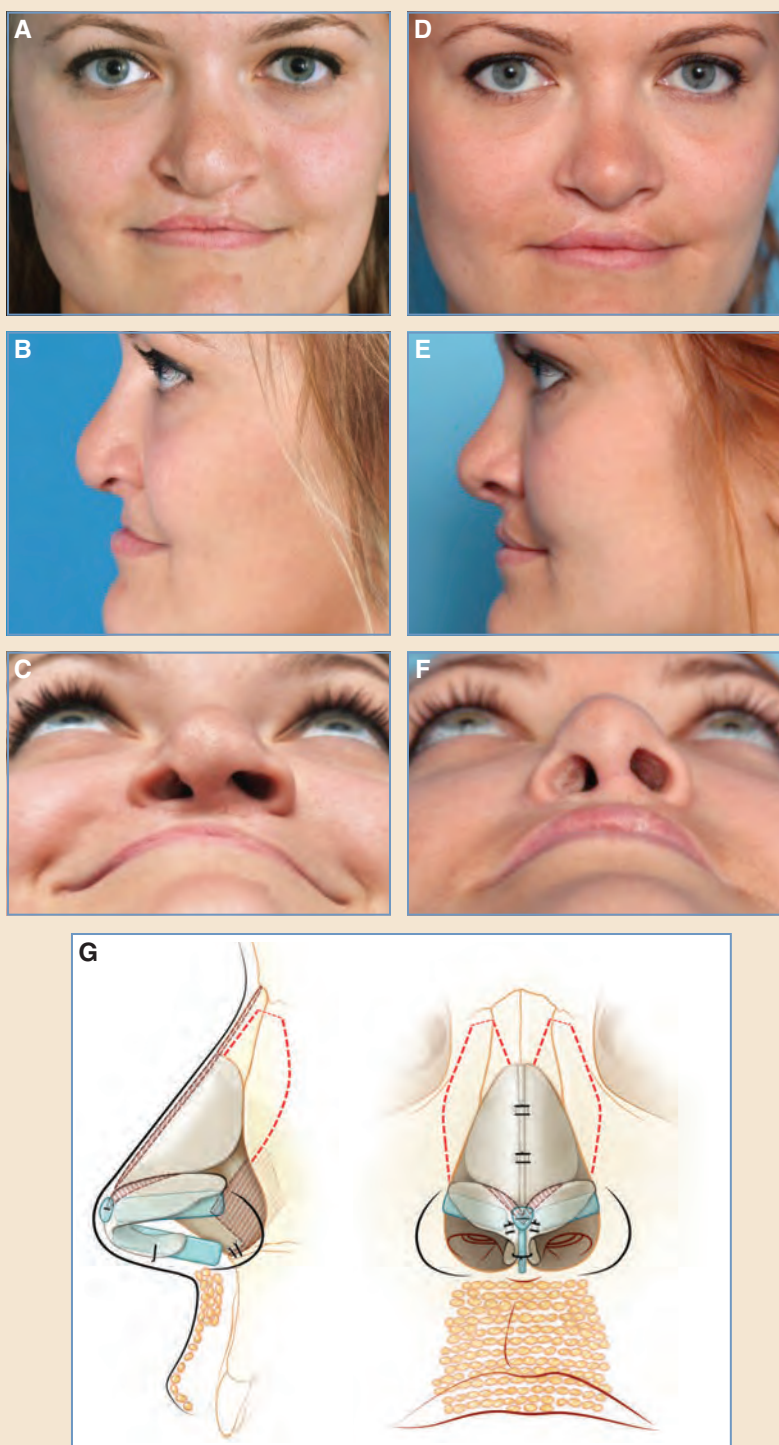


Fig. 21-27 This bilateral cleft patient underwent septorhinoplasty using an open approach with bilateral extended columellar incisions, as described previously. **A-C**, Preoperative views. **D-F**, One-year postoperative views. **G**, A secondary septoplasty with cartilage excision and caudal strut repositioning was performed. Rib graft was harvested, given the lack of native septal cartilage. The tip was reconstructed with bilateral alar strut grafts (both lower lateral cartilage–alar strut grafts were repositioned to a more inferior pocket), a columellar strut, interdomal and intradomal sutures, tip grafting, and cephalic trim. Bilateral medial and lateral osteotomies were performed. Fat was grafted to the nasolabial area and upper lip.

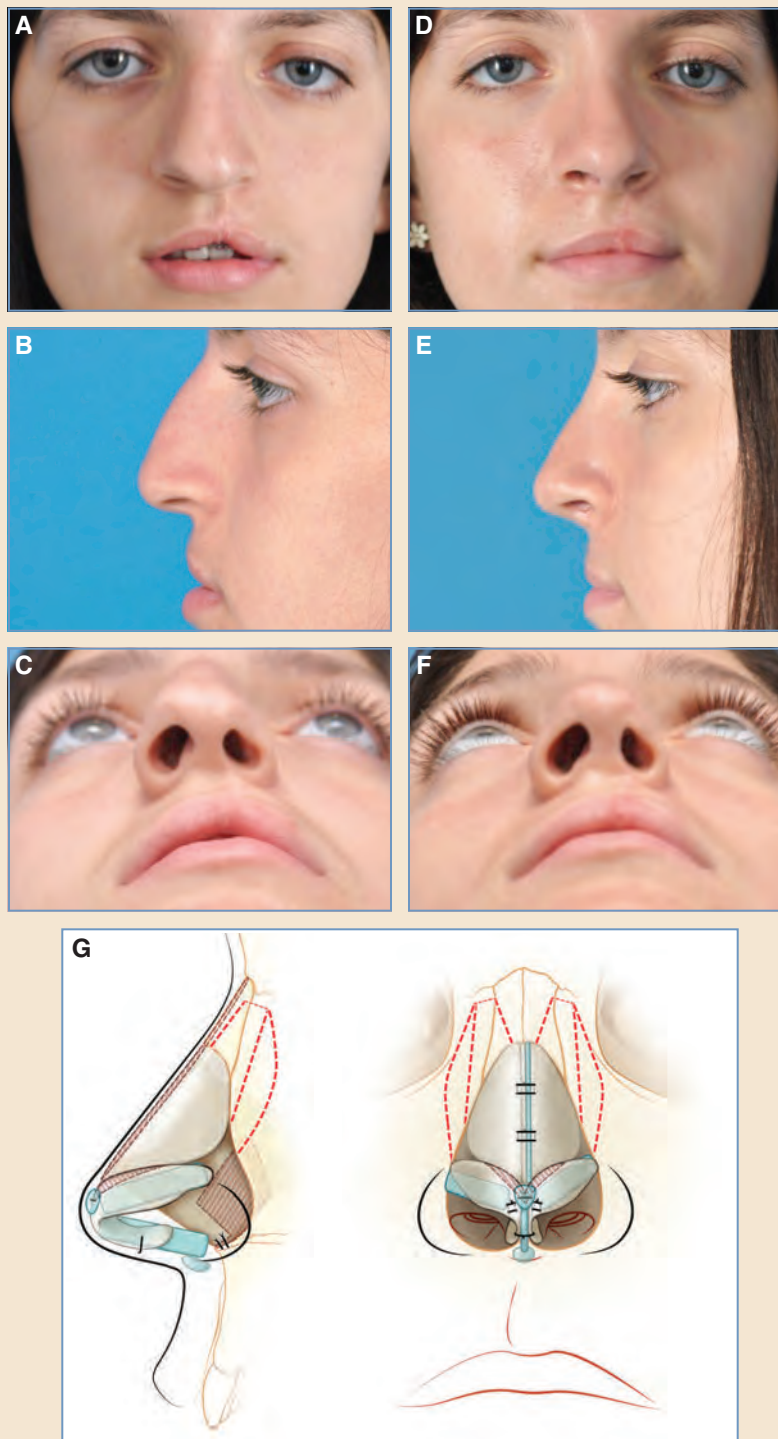


Fig. 21-28 This unilateral cleft patient underwent septorhinoplasty using an open approach with the extended columellar incision described previously. **A-C**, Preoperative views. **D-F**, One-year post-operative views. **G**, Septoplasty was performed with submucous resection and scoring. The tip was reconstructed with bilateral alar strut grafts (the right lower lateral crus was repositioned inferiorly with a strut graft), a columellar strut, tip graft, nasolabial angle graft, interdomal and intradomal sutures, and cephalic trim. The dorsum was rasped. Bilateral medial and double-lateral osteotomies were performed. A spreader graft was placed along the left side.

OTHER CONGENITAL NASAL DEFORMITIES

Other congenital, noncleft-related deformities can occur that may require the techniques discussed in this chapter. Median clefts and dermoid lesions can result in a widened nose, malformed septum, and/or bifid septum. These nonspecific congenital deformities may present myriad challenges. As mentioned previously, regardless of the deformity, rhinoplasty surgeons can use the techniques presented to do whatever is needed to make the nose look normal.

CONCLUSION

The artistry of cleft repair continues to evolve. With the shared experience of centuries of cleft surgery, more aesthetic and functional results can be accomplished with primary lip and nose surgery. However, despite our best efforts to reduce the stigmata of the primary deformity, secondary deformities occur, and secondary correction is usually required. The repair of primary nasal deformities requires patience. A gradual, logical approach directed at staged correction of the primary deformities, without introducing iatrogenic secondary deformities, is encouraged.

KEY POINTS

- Secondary deformities of the lip and nose arise because of limited surgical correction at the initial repair and because key anatomic landmarks were not identified and aligned.
- Although each surgeon has a preferred technique for lip repair, the anatomic subunit principles described by Fisher can be similarly applied to correction of secondary deformities (see Chapter 19).
- Understanding Noordhoff's point and its importance in the design of the lateral vermilion muscular flap to fill a medial vermilion defect is essential to prevent vermilion deficiency and a whistle deformity.
- In all but the very minor forms of unilateral cleft lip, the vermilion below the cleft side of the bow is vertically deficient. If this is not augmented with vermilion from the lateral lip element during the primary repair, a stairstep deformity of the vermilion-mucosal junction (red line) will be present.
- A major cleft lip revision offers an opportunity to reposition a scar to better mirror the noncleft-side philtral column and, for this reason, may be preferable to a minor revision.
- The surgical goals of a major lip revision are similar to those of a primary repair; atypical secondary deformities may need to be addressed using the age-old principles of re-creating the defect and anatomically reconstructing the lip.

Continued

KEY POINTS (continued)

- The degree of secondary nasal deformity depends on the degree of primary nasal deformity and the extent of primary rhinoplasty.
- An analysis of a secondary nasal deformity involves a stepwise assessment of the uncorrected primary deformities.
- The goals of a secondary cleft rhinoplasty are to create a symmetrical, normal- and natural-looking nose and to optimize breathing and function. Although a primary cleft rhinoplasty can do much to achieve a more normal-looking nose, the limitations of dissection and bone and septal work can lead to secondary deformity as a patient enters the teenage years.
- No matter what deformities are encountered during a definitive cleft rhinoplasty, the goal should be to do whatever it takes to make the tip structures look normal.
- A Tajima reverse-U procedure and its modifications can be applied both in primary bilateral cleft lip nasal repair for columellar lengthening and in early and late secondary correction of a unilateral cleft nasal deformity, with the incision placement lending itself well to late, open rhinoplasty.
- A definitive septorhinoplasty should ideally be delayed until skeletal maturity, but earlier correction is warranted in some cases and can be safely performed.
- The most difficult secondary deformities to correct are of iatrogenic origin.

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22

Cleft Palate Deformities

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he existence of a facial cleft in a child requires long-term treatment planning. Because facial clefts affect both function and facial aesthetics, therapy and rehabilitation should begin soon after birth and may continue for many patients until the late teens or early twenties. Although functional effects range from early feeding difficulties to growth attenuation and secondary effects on middle ear function, by far the most significant effect is on speech. The disturbance is typically in the form of *hypernasality* or escape of sound through the nasal cavity on production of many consonants, phonemes, and vowels in the English language. The importance of speech for normal social functioning cannot be underestimated.

Many surgical techniques and modifications have been advocated to improve functional outcome and aesthetic results. The most controversial issues in the management of cleft palate are the timing of surgical intervention, the effects of various surgical procedures on the development of speech, and the effects of surgery on facial growth. We will address these issues in this chapter by reviewing the various techniques and discussing our current preferences and why these procedures are preferred.

EMBRYOLOGY

The primary palate is the keystone to the upper lip and anterior portion of the definitive palate. Its embryogenesis is fundamental to normal development of the midface, and its maldevelopment has profound clinical and sociologic consequences with regard to breathing, sucking, swallowing, mastication, osculation, speech, and facial physiognomy.

Orofacial development in the embryo is first demarcated by the appearance of the prechordal plate at the cranial end of the embryonic disc at postconception day 14.¹ This plate designates the future mouth, or stomodeum. The mesenchyme that provides the facial primordia is oddly of ectodermal derivation, arising from neural crest cells at the apices (crests) of the neural folds

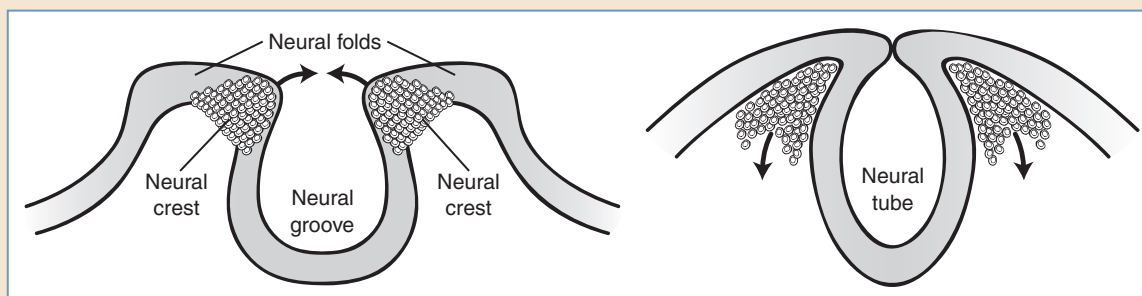


Fig. 22-1 Neural crest origin and migration.

before neural tube formation² (Fig. 22-1). The neural crest cells oddly disrupt the ectodermal-mesodermal boundary and migrate into the subjacent tissue as ectomesenchymal cells. Their migration and proliferation are fundamental to facial development. During their migration they interact with the extracellular matrix and adjacent epithelia, which partly determine the patterning and nature of the derivative tissues they will form. These derivatives include neural, skeletal, connective, and muscular tissues.³ Migration of neural crest cells into the five pharyngeal arches, which they in part create, occurs in a highly regulated manner, under the control of homeobox genes.⁴

The stomodeal chamber, which is the precursor of the future mouth, becomes a deepened central depression in the facial region as a consequence of the surrounding five primordia of the face, bulging at the borders of the stomodeum (Fig. 22-2). The *five primordia* are the single median and rostrally located frontonasal prominence and the paired bilateral maxillary and caudally located mandibular prominences.⁵ These prominences are the result of the migration and mitotic proliferation of neural crest ectomesenchyme, which originates from the caudal region of the mesencephalon and the rhombencephalon of the developing brain. There is also a mesodermal contribution to the mesenchyme of the facial prominences.

Formation of the upper lip involves an extraordinary combination of elements arising from both the frontonasal prominence and maxillary prominence, which are components of the first pharyngeal or mandibular arch. The bilateral medial nasal prominences form the central tuberculum of the upper lip and provide the basis for the primary palate.^{6,7} The upper lip is completed on either side of the central tuberculum by fusion of the freely projecting medial nasal prominences with the laterally located maxillary prominences, requiring critically timed correlation of their growth, spatial location, and disintegration of their contacting surface epithelia that form the transient nasal fin.⁸ Disintegration of the nasal fin by apoptosis or mesenchymal transformation allows intermingling of the underlying mesenchymal cells, providing continuity to the median and lateral components of the upper lip. Failure of normal disintegration of the nasal fin or inadequate mesenchymal migration across the two boundaries of the maxillary medial nasal prominences results in lip clefting, unilaterally or bilaterally, with varying degrees of severity.

Fusion of the medial nasal and maxillary prominences not only provides continuity of the upper jaw and lip but also separates the nasal pits from the stomodeum. The central median component of the face forms the tuberculum and philtrum of the upper lip, the tip of the nose, and

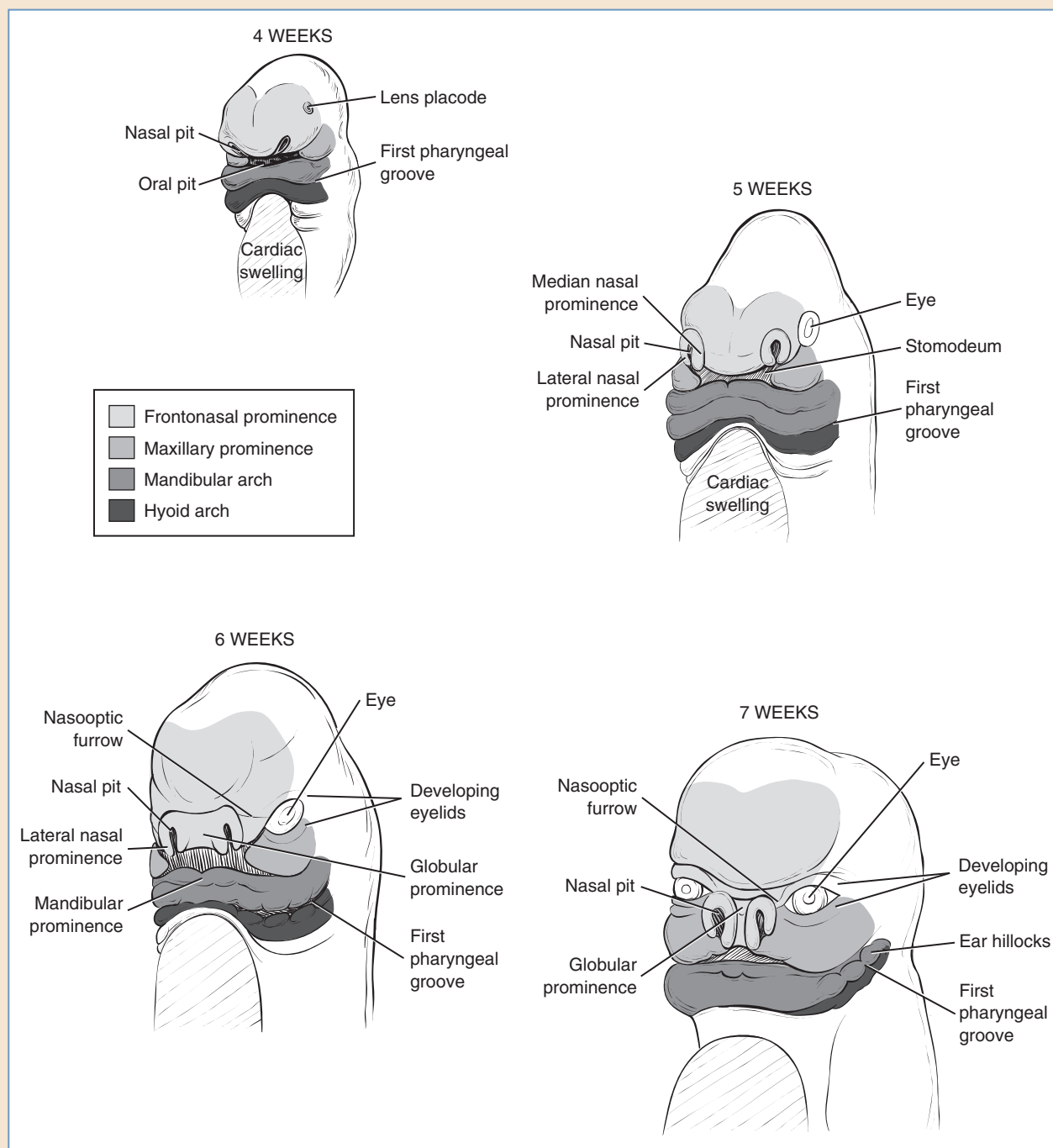


Fig. 22-2 Facial formation from 4 to 7 weeks postconception.

the primary palate. The intermaxillary segment of the upper jaw, the premaxilla, in which the four upper incisor teeth will develop, arises from the medial primary palate.⁹ The rare absence of the primary palate is demonstrated in *premaxillary agenesis*, producing a median cleft lip and palate (CL/P)—a manifestation of holoprosencephaly.¹⁰

Normally four tooth buds, which produce the upper incisor teeth, arise in the primary palate. The location of these four teeth, which are demarcated by incisive fissures in the fetus, usually defines the limits of the primary palate. The canine teeth normally arise in the secondary palate. Most commonly, clefting between the primary and secondary palates occurs at the incisive fissure that separates the lateral incisor and canine teeth.

The *secondary palate*, so called because it forms after the appearance of the primary palate, constitutes both the floor of the nasal cavities and the roof of the mouth. The *secondary palate* is composed of the anterior hard palate and posterior soft palate, and it is an essential component of normal respiration, mastication, deglutition, and speech.

Three elements make up the secondary definitive palate: the two lateral palatal processes projecting into the stomodeum from the maxillary prominences and the primary palate derived anteriorly from the frontonasal prominence. These three elements are initially widely separated as a result of the advancing edges of the palatal processes being deflected down on either side of the protruding tongue, which initially occupies most of the stomodeal chamber.¹¹ Concomitantly, the midline cartilaginous nasal septum descends from the roof of the stomodeum as a feature of nasal capsular development.¹² During postconception week 8, a remarkable transformation of the palatal shelves occurs when they elevate into a horizontal position as a prelude to their fusion with each other, the primary palate, and the nasal septum, thereby partitioning the oronasal chamber.

The transition from vertical to horizontal is completed within hours during postconception week 8. There is a sex difference in the timing of palatal closure; shelf elevation and fusion begin a few days earlier in male embryos than in female embryos¹³; the slight delay possibly accounts for the higher incidence of cleft palates in females. Several mechanisms have been proposed for the rapid elevation of the palatal shelves. They include biochemical transformations in the physical consistency of the connective tissue matrix of the shelves; variations in vasculature and blood flow to these structures, resulting in a sudden increase in tissue fluid turgor; rapid differential mitotic growth; muscular movements; and an intrinsic shelf force.^{14,15} The intrinsic shelf elevating force is generated chiefly by the synthesis, accumulation, and hydration of hyaluronic acid and glycosaminoglycans within the extracellular matrix of the shelves.¹⁶

The withdrawal of the fetus' face from against the heart prominence by uprighting the head facilitates jaw opening. Mouth-opening reflexes and extrinsic tongue muscle activity have been implicated in the withdrawal of the tongue from between the vertical shelves.¹⁷ Depressed fetal swallowing may delay palatal shelf elevation, precluding their conjunction and leading to clefting. Prenatal ultrasonography revealing aphagia has been correlated with cleft palate formation.¹⁸ The fetus must be in a floating position in the amniotic sac to permit jaw movements; in the event of oligohydramnios, deficient amniotic fluid inhibits mouth opening and tongue withdrawal, thereby precluding palatal shelf elevation and thus accounting for a possible source of clefting.

During palate closure the mandible becomes more prognathic and the vertical dimension of the stomodeal chamber increases, although maxillary width remains stable, allowing shelf contact to occur. Also, forward growth of Meckel's cartilage relocates the tongue more anteriorly, concomitant with head elevation. Mandibular growth retardation causes *retrognathia*, which enforces a high tongue position and prevents the shelves from fusing—a series of developmental malformations that lead ultimately to the Pierre Robin sequence.¹⁹

Ossification of the palate proceeds during postconception week 8 from the spread of bone into the mesenchyme of the fused lateral palatal shelves and from trabeculae appearing in the primary palate as premaxillary centers, all derived from the single primary ossification centers of the maxillas.²⁰⁻²² Posteriorly, the hard palate is ossified by trabeculae spreading from the single primary ossification center of each of the palatine bones.

Ossification does not occur in the most posterior part of the palate, giving rise to the soft palate. Myogenic mesenchymal tissue of the first and fourth pharyngeal arches migrates into this faucial region, supplying the musculature of the soft palate and fauces.²³ The tensor veli palatini is derived from the first pharyngeal arch; the levator palatini and uvular and faucial pillar muscles are derived from the fourth pharyngeal arch, accounting for the innervation by the first arch trigeminal nerve of the tensor veli palatini muscle and by the fourth arch pharyngeal plexus and vagus nerves for all of the other muscles.

Fusion of the three palatal components initially produces a flat, unarched roof to the mouth. The hard palate grows in length, breadth, and height, becoming an arched roof for the mouth. The fetal palate increases in length more rapidly than in width between 7 and 18 weeks after conception, after which the width increases faster than the length.²⁴ At birth the length and the breadth of the hard palate are almost equal. The postnatal increase in palatal length is the result of appositional growth in the maxillary tuberosity region and, to some extent, at the transverse maxillopalatine suture.²⁵

Anomalies of the palate occur as a consequence of disturbances to the developmental processes. Successful fusion of the three embryonic components of the palate involves complicated synchronization of shelf movements with growth and withdrawal of the tongue and growth of the mandible and head. Mistiming of any of these critical events, because of environmental agents or genetic predisposition, results in failure of fusion, leading to clefts of the palate.

FREQUENCY

Cleft palate can occur as an isolated cleft palate or in combination with a cleft lip. The isolated cleft palate is genetically and morphologically different from a CL/P. The former has an incidence of 0.45 to 0.5 in 1000 births and is equal across all ethnic backgrounds. On the other hand, the incidence of cleft lip with cleft palate by race is 2.1 in 1000 in Asians, 1 in 1000 in whites, and 0.41 in 1000 in blacks. A large surveillance data-based study recently published²⁶ showed a significant trend toward an increasing prevalence of cleft palate in the United States alone. These data, however, remain under debate, because there is no uniform registration of clefts worldwide. The foremost type of clefting is a *bifid uvula*, which occurs in 2% of the population. The second most frequent type is a *left unilateral complete cleft of the palate and prepalatal structures*. Midline clefts of the soft palate and parts of the hard palate are also common. Complete clefts of the secondary palate are twice as common in females as in males, whereas the reverse is true of velar clefts. Isolated cleft palate generally has a much higher incidence of associated anomalies, resulting in diagnosis of a syndrome in almost one third to one half of patients.²⁷ This is distinctly different from CL/P, in which it is unusual to have other associated anomalies or an identifiable syndrome.

Recurrence risks for cleft palate deformities do not correspond to any Mendelian pattern of inheritance, and it appears that clefting is inherited heterogeneously. This observation is supported by evidence from studies of twins that indicate the relative roles of genetic and nongenetic influences of cleft development. For isolated cleft palate and combined CL/P, if the proband has no other affected first-degree or second-degree relatives, the empiric risk of a sibling being born

with a similar malformation is 3% to 5%. However, if a proband with a combined CL/P has other affected first-degree relatives, the risk for siblings or subsequent offspring is 10% to 20%.

CAUSE

Nonsyndromic clefting of the CL/P in humans has a highly complex etiology, with both multiple genetic loci and exposure to teratogens influencing individual susceptibility.²⁸ The familial clustering of CL/P has been extensively characterized, and epidemiologic studies have proposed monogenic models (putative major locus associated with reduced penetrance),²⁹ multifactorial threshold models, and mixed major gene/multifactorial models to explain its inheritance.²⁹⁻³¹ Clefting can result from a single gene defect either as part of a syndrome (for example, Van der Woude syndrome, Treacher Collins syndrome, velocardiofacial syndrome, Apert syndrome, and other fibroblast growth factor receptor 2 [FGFR2]–mediated craniosynostosis syndromes³²) or as an isolated phenotypic effect (for example, X-linked cleft palate and nonsyndromic, autosomal dominant orofacial clefting). Several studies have suggested that chromosome 6p is a candidate region for a locus involved in orofacial clefting,³³ and linkage studies suggest another susceptibility locus on the long arm of chromosome 4.³⁴ Although evidence of the involvement of a major gene as the cause of CL/P has been reported, orofacial clefting is genetically complex, with no single gene responsible for all forms. Molecular studies provide further insight into the genetic mechanism underlying CL/P, and these findings have important implications with regard to the feasibility of detecting linkage to loci, conferring susceptibility to CL/P.³⁵

Genetic analysis and tissue-specific expression studies support a role for transforming growth factor alpha (TGF- α) in craniofacial development. Numerous studies have confirmed an association at the TGF- α locus with nonsyndromic CL/P in humans,^{30,36} extending the role of TGF- α in craniofacial morphogenesis and supporting an interrelated mechanism for this major gene as the cause of nonsyndromic forms of CL/P.³⁷ In addition, a gene-environment interaction between maternal smoking, TGF- α , and clefting was reported. Shaw et al³⁸ offered evidence that the risk for orofacial clefting in infants may be influenced by maternal smoke exposure alone and in combination (gene-environment interaction) with the presence of the uncommon TGF- α allele. This was supported in a study by Hwang et al,³⁹ in which infants with isolated birth defects (CL/P and control subjects with noncleft birth defects) were tested for an association among maternal exposures, genetic markers, and oral clefts. A modest increase in the less common C2 allele of the TGF- α locus was seen among infants with cleft palate only compared with control subjects with other birth defects, and the association appeared to reflect an underlying interaction between maternal smoking and infant genotype. This apparent gene-environment interaction was also found in those reporting no family history of any birth defects.

Even though the first association studies of CL/P with candidate genes found an association with TGF- α , other candidate genes have also been identified. An association of CL/P with the retinoic acid receptor alpha locus has been reported,³⁵ as well as associations with the *BCL-2 oncogene*, *BCL-3*,⁴⁰ *Sonic hedgehog*, *paired box gene 9* (PAX9),⁴¹ epidermal growth factor receptor,⁴² and several of the known homeobox genes.⁴³ Although some studies suggest that the familial aggregation of nonsyndromic CL/P is likely attributable to the effect of several susceptibility loci acting in a multiplicative fashion,³⁵ others report no interaction between the TGF- α and retinoic acid receptor alpha polymorphisms, even though jointly they appear to account for almost half of the attributable risk of clefting.⁴³

Another gene identified as having a potential role in clefting is transforming growth factor beta 3 (TGF-beta 3). Kaartinen et al,⁴⁴ using TGF-beta 3 null mutant mice, showed an essential function for this gene in normal morphogenesis of the palate, directly implicating a role for this gene in the epithelial-mesenchymal transformation necessary for palatal fusion. Mice lacking TGF-beta 3 had an incompletely penetrant failure of the palatal shelves to fuse, thus leading to cleft palate. In addition to the identification of major genes involved in the cause of nonsyndromic CL/P, information through the human genome project and the use of animal models have facilitated the study of gene environment interactions with respect to orofacial clefting. For example, an interaction has been shown between maternal periconceptual health and the TGF-alpha polymorphism, leading to an increased risk for CL/P.³⁸ In addition, it has been reported that maternal multivitamin supplementation may lead to a reduced risk of CL/P in infants having the TGF-alpha mutation.⁴⁵ Pharmaceutical compounds, such as anticonvulsant drugs,⁴⁶ corticosteroids,⁴⁷ folate antagonists, and retinoids,⁴⁸ as well as alcohol, maternal hyperthermia, and organic solvents/agricultural chemicals, have been shown to cause CL/P in exposed offspring (see following section). However, the mechanism by which many of these compounds exert their teratogenic effect remains unknown. Recent technologic advances, however, will allow the analysis of critical changes in gene expression subsequent to teratogen exposure and will promote the discovery of genetic polymorphisms that will enhance our ability to identify individuals at risk.

Environmental Risk Factors and Oral Clefts

Although the literature regarding the cause of oral clefts is extensive, unique causal factors remain unknown. The widely held belief is that oral clefts are of multifactorial origin, with both genetic predisposition and environmental influence playing a role. The term *environmental* is used in its broadest sense to include characteristics of the individual, such as age, sex, and race, parental behaviors such as smoking and alcohol use, and other types of exposures such as occupational or environmental. Although no strong risk factors for oral clefts have been identified, several potential environmental risk factors have been investigated. Box 22-1 summarizes the major environmental risk factors associated with oral clefts. The association is weak for most risk factors and inconclusive for others.⁴⁹ Most associations are stronger for CL/P rather than for isolated cleft palate.

Box 22-1 Environmental Risk Factors Associated With Oral Clefts

- Maternal cigarette smoking^{38,50-55}
- Alcohol consumption⁵⁶⁻⁵⁹
- Caffeine⁶⁰
- Epilepsy and anticonvulsants⁶¹⁻⁷⁰
- Benzodiazepines⁷¹⁻⁷⁵
- Corticosteroids⁷⁶⁻⁸⁰
- Organic solvents/pesticides⁸¹⁻⁸²
- Dioxins⁸³
- Maternal nutrition⁸⁴:
 - Folic acid⁸⁵⁻⁸⁶
 - Vitamin B₆⁸⁷⁻⁸⁸
 - Vitamin A⁸⁹⁻⁹¹

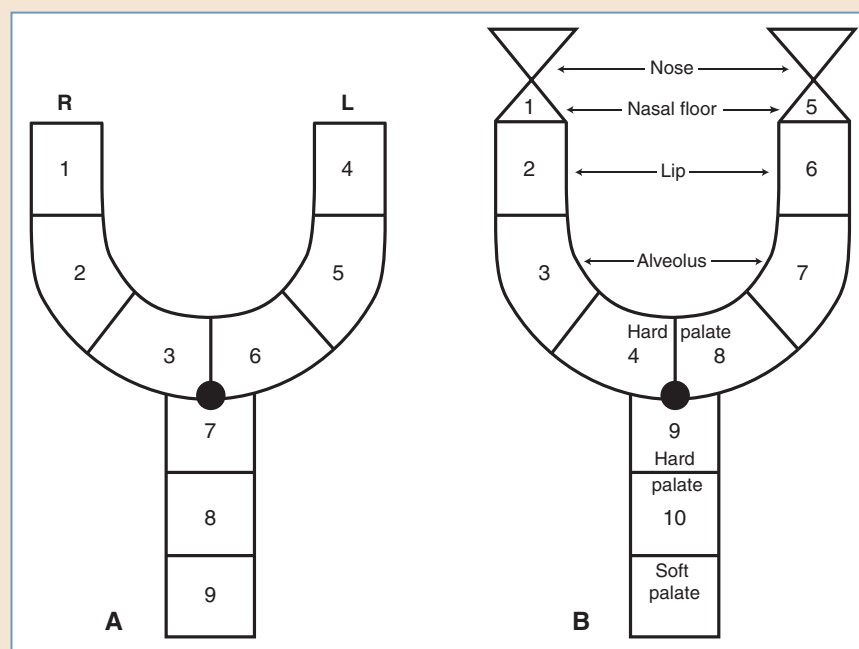


Fig. 22-3 Symbolic representation of cleft lip and palate. **A**, Classic Kernahan "Y." **B**, Millard's modification.

CLASSIFICATION OF DEFECT

It is important to accurately analyze and report a cleft lip and/or palate deformity in a standardized manner. This enables optimal analysis and perioperative planning, and it facilitates multi-center reporting and analysis both retrospectively and prospectively. A classification system presented by the American Association for Cleft Palate Rehabilitation was originally published in 1962, and it was later accepted by the Cleft Palate Association. However, because of its complexity, it was not uniformly accepted. In 1971 Kernahan introduced a simpler classification scheme that may be reported on a diagrammatic Y-shaped symbol, with the incisive foramen represented at the focal point⁹² (Fig. 22-3). This was subsequently used by Millard, and other versions were later proposed to allow easier computerized entry for all anatomic cleft variants.⁹²

PERTINENT NORMAL AND ABNORMAL ANATOMY

The bony portion of the palate is a symmetrical structure with division based on the embryonic origin into the primary and secondary palate. The premaxilla, alveolus, and lip, which are anterior to the incisive foramen, are parts of the primary palate. Structures posterior to it, which include the paired maxilla, palatine bones, and pterygoid plates, are part of the secondary palate. The severity of the clefting of the bony palate varies from simple notching of the hard palate to clefting of the alveolus.

The palatine bone is located posterior to the maxilla and pterygoid lamina. It is composed of horizontal and pyramidal processes. The horizontal processes contribute to the posterior aspect of the hard palate and become the floor of the choana. The pyramidal processes extend vertically to contribute to the floor of the orbit.⁹³

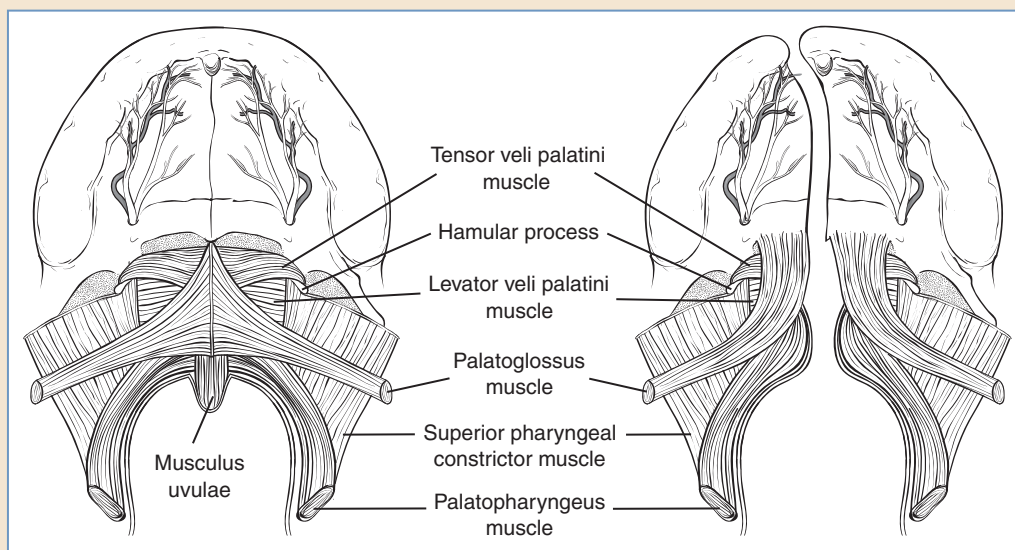


Fig. 22-4 Muscular attachments to the palate.

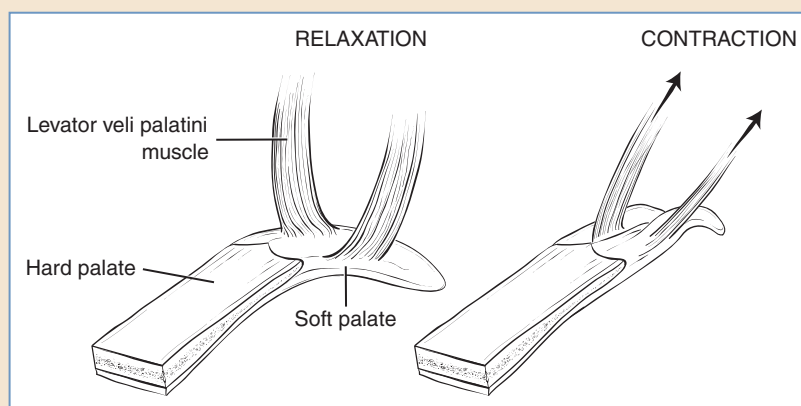


Fig. 22-5 The effect of the contracting arms of two levator palatini muscles.

Even though the bony defect is important in the surgical treatment of cleft palate, the pathology in the muscles and soft tissues has the greatest impact on the functional result. Six muscles are attached to the palate: the levator veli palatini, superior pharyngeal constrictor, musculus uvulae, palatopharyngeus, palatoglossus, and tensor veli palatini (Fig. 22-4). The three muscles that appear to have the greatest contribution to velopharyngeal function are the levator veli palatini, musculus uvulae, and superior pharyngeal constrictor. The musculus uvulae muscle acts by increasing the bulk of the velum during muscular contraction.^{94,95} The levator veli palatine pulls the velum superiorly and posteriorly to oppose the velum against the posterior pharyngeal wall (Fig. 22-5). The medial movement of the pharyngeal wall, attributed to the superior pharyngeal constrictor, aids in the opposition of the velum against the posterior pharyngeal wall to form the competent sphincter. The palatopharyngeus displaces the palate downward and medially.

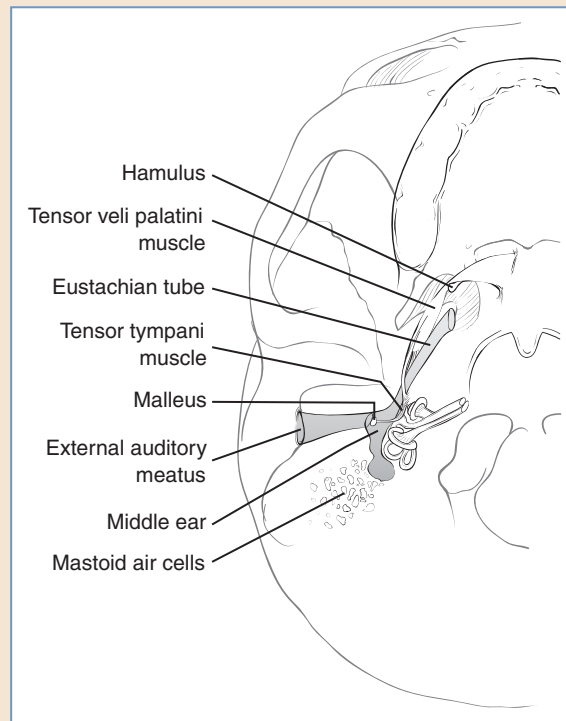


Fig. 22-6 The function of the tensor veli palatini is to improve the ventilation and drainage of the auditory tubes.

The *palatoglossus* is mainly a palatal depressor that plays a role in the production of phonemes with nasal coupling by allowing controlled airflow into the nasal chamber. The tensor veli palatini does not contribute to the movement of the velum. The tendons of the tensor veli palatini hook around the hamulus of the pterygoid plates, and the aponeurosis of the muscle inserts along the posterior border of the hard palate. The muscle originates partially on the cartilaginous border of the auditory tubes. The function of the tensor *veli palatini*, similar to the *tensor tympani*, with which it shares similar innervation, is to improve the ventilation and drainage of the auditory tubes (Fig. 22-6).

In cleft palate, the aponeurosis of the tensor veli palatini, instead of attaching along the posterior border of the hard palate, is attached along the bony cleft edges. All the muscles that attach to the palate insert onto the aponeurosis of this muscle. Thus the overall length of the palate is shortened. The abnormality in the tensor veli palatini increases the incidence of middle ear effusion and infection.

The muscle sling of the levator veli palatini is also interrupted by cleft palate. The levator does not form a complete sling. The medial portion of each side attaches to the medial edge of the hard palate. Thus in patients with cleft palate, the effectiveness of the velar pull against the posterior pharyngeal wall is impaired. Of the six muscles, the prevailing theory considers the contribution of the levator veli palatini to be the most important factor in establishing velopharyngeal competence.⁹⁶

PATHOPHYSIOLOGY

The pathologic sequelae of cleft palate include feeding and nutritional difficulties, recurrent ear infections, hearing loss, abnormal speech development, and facial growth distortion. The abnormal communication between the oral and nasal chamber impairs the normal sucking and swallowing mechanism of the cleft infant. Food particles often reflux into the nasal chamber. The abnormal insertion of the tensor veli palatini prevents satisfactory emptying of the middle ear. Recurrent ear infections have been implicated in the hearing loss of patients with cleft palate, and hearing loss may worsen the speech pathology in these patients.

Evidence that repair of the cleft palate decreases the incidence of middle ear effusions is inconsistent; however, these problems are overshadowed by the magnitude of speech and facial growth problems. Speech abnormalities are intrinsic to the anatomic derangement of cleft palate. The facial growth distortion appears to be, to a great extent, because of surgical interventions. An intact velopharyngeal mechanism is essential for the production of nonnasal sounds, and it is a modulator of the airflow in the production of other phonemes that require nasal coupling.⁹⁷ The complex and delicate anatomic manipulation of the velopharyngeal mechanism, if not successfully learned during early speech development, can permanently impair normal speech acquisition.⁹⁸

The closure of the velopharyngeal sphincter prevents the flow of air into the nasal chamber during the production of specific phonemes. This mechanism is essential in the production of nonnasal sounds. Speech impairments specific to velopharyngeal dysfunction include sibilant consonants: the sounds *s*, *z*, *sh*, *zh*, and *ch*; *j* or soft *g* and the intraoral pressure consonants *p*, *b*, *t*, *d*, *k*, and hard *g*. The brain responds to the inability to close the velopharynx by adopting compensatory articulation patterns, which unfortunately are maladaptive and can make speech completely unintelligible. Speech therapy is usually indicated for children with CL/P, generally beginning in the first 2 years and continuing until normal articulation is established.

Multiple studies have demonstrated that the cleft palate maxilla has some intrinsic deficiency of growth potential. The degree of intrinsic growth potential deficiency varies from isolated cleft of the palate to complete CL/P. Growth potential is further impaired by surgical repair. Any surgical intervention performed before the completion of full facial growth can have significant deleterious effects on maxillary growth. Disagreement exists regarding the appropriate timing of surgery to minimize the harmful effects on facial growth and what type of surgical intervention is most responsible for growth impairment. The formation of scar and scar contracture in the areas of denuded palatal bones is most frequently blamed for restriction of maxillary expansion.

The growth disturbance is exhibited most prominently in the prognathic appearance during the second decade of life despite the normal appearance in early childhood. The discrepant occlusal relationship between the maxilla and mandible is usually not amenable to nonsurgical correction.

THE HISTORY OF CLEFT PALATE SURGERY

Rogers expertly reviewed the evolution of cleft palate surgery.^{99,100} The first record of a palatal operation dates back to 500 AD, and it was prompted by inflammation of the uvula. In 1552, Houlier proposed suturing palatal clefts,¹⁰⁰ and 12 years later Ambroise Paré¹⁰¹ illustrated obturators for palatal perforation. In 1764, Le Monnier, a French dentist, successfully repaired a cleft velum with a few sutures and hot cautery of the edges.¹⁰² Von Graefe, 50 years later, produced inflammation of the velar margins before bringing them together in his palate suture, and he is credited with performing the first velar repair of a cleft in 1816.¹⁰³ J.C. Warren¹⁰⁴ performed the first velar closure in America in 1824.

In 1828, Dieffenbach¹⁰⁵ enhanced the surgical treatment of cleft palate by introducing hard palatal mucosa elevation to allow closure of the hard palatal cleft; in 1861 von Langenbeck¹⁰⁶ proposed the creation of a bipediced mucoperiosteal flap that could be mobilized medially to close the palatal cleft. The improved vascular supply to the mucoperiosteal flap significantly decreased the incidence of dehiscence.

The ability to successfully close the palate led to concern about palatal function. It was evident by this time that the short and immobile palate impaired the speech capability of patients with cleft palate. Veau,¹⁰⁷ Kilner,¹⁰⁸ and Wardill¹⁰⁹ described a repair that uses four flaps to accomplish the palatal closure. Two anterior palatal flaps close off the region of the incisive foramen while the posterior palatal flaps are pushed back to lengthen the palate.¹¹⁰

Schweckendiek¹¹¹ advocated the use of a two-stage cleft palate closure. The soft palate was closed early, with closure of the hard palate delayed until several years later. The rationale for the two-stage procedure was to provide improved velopharyngeal function during initial speech development and to accomplish the closure of the hard palate after the cleft narrows with facial growth. Anatomic muscle realignment has also been postulated as essential in improving postoperative velopharyngeal function. These historic developments underlie the existing controversies in management of cleft palate.

Modern Era of Palate Repair

In the late 1950s Randall¹¹² refined the technique originally described by Veau, Wardill, and Kilner. Later, Bardach et al¹¹³ introduced the concept of two-flap palatoplasty, which further improved mobility for coverage of the anterior cleft. Then, in 1986, Furlow¹¹⁴ described his double-opposing Z-plasty technique for palatal repair, which will be described in this chapter.

PLANNING AND TIMING OF PALATAL CLOSURE

The goal of cleft palate treatment is to separate the oral and nasal cavities. Although it is not absolutely necessary for feeding, it is advantageous to normalize feeding and decrease regurgitation and nasal irritation. More important than repairing the oral and nasal mucosa is the repositioning of the soft palate musculature to anatomically re-create the palate so that normal speech can be established. Another goal of palate repair is to minimize restriction of growth of the maxilla in both sagittal and transverse dimensions.

The goals of palatal repair include normal speech, normal palatal and facial growth, and normal dental occlusion.¹¹⁵⁻¹¹⁷ Physicians believe that early palatal repair is associated with better speech results, but early repair also tends to produce more severe dentofacial deformities.¹¹⁸⁻¹²⁰ Randall et al,¹²¹ as well as Lehman and colleagues,¹²² consistently reported that children whose palates were repaired at an earlier age appeared to have better speech and needed fewer secondary pharyngoplasties than those whose surgeries had been delayed beyond the first 12 months.

Noordhoff and associates¹²³ found that children undergoing delayed palatoplasty for cleft palate had significantly poorer articulation skills before the hard palate closure than children of the same age who did not have clefts. These benefits of early cleft palate repair from the standpoint of speech and hearing must be weighed against the increased technical difficulty of the procedure at a younger age and possible adverse effects on maxillary growth.

In our institutions, palatoplasty is performed when the patient is 6 to 12 months of age, depending on the extent of the cleft. Incomplete clefts can be repaired earlier than complete clefts. Surgery at this age results in improved speech development. This protocol is supported by the fact that numerous studies failed to demonstrate an observable difference in underdevelopment of the palatal arch among children undergoing operations at various ages. The surgical intervention

appears to interfere with midfacial growth regardless of the age of the patient at the time surgery is performed.¹²⁴ Patients with Pierre Robin sequence and those with idiopathic micrognathia typically undergo repair at 12 to 13 months of age unless the airway is severely compromised. Some plastic surgeons (Parri and others¹²⁵) advocate early soft palate repair at 3 to 6 months and repair of the remaining cleft by 1 year, although this is not our approach.

SURGICAL TECHNIQUE

Numerous techniques exist for surgical repair of the cleft palate. In general, operations are designed to achieve two primary goals: restoration of hard palate continuity and reconstruction of normal soft palate anatomy and function. The flaps most commonly used for hard palate continuity are bipedicle mucoperiosteal flaps, such as in the von Langenbeck repair, or axial mucoperiosteal flaps based on a single pedicle of the greater palatine vessels, as proposed in the Veau-Wardill-Kilner “pushback” and the Salzer-Bardach two-flap repair.¹²⁶ Vomer flaps may be required to assist in closure of the nasal lining in wide and bilateral clefts. In very wide clefts or in secondary repairs, some authors advocate the use of a calvarial periosteal graft,¹²⁷ buccal myomucosal flap,¹²⁸ acellular dermal matrix,¹²⁹ or free tissue transfer.^{130,131} Another interesting concept in the treatment of wide clefts, which is the focus of ongoing research, is the use of palatal tissue expansion.^{132,133} Black and Gampper¹³⁴ suggested the use of a rotational rather than the traditional advancement mucoperiosteal flap, allowing off-midline closure. However, all the above-mentioned newly developed modalities lack the long-term follow-up necessary for establishing a routine surgical protocol.

Procedures used to re-create the normal anatomy of the soft palate include intravelar veloplasty and the Furlow or double-opposing Z-plasty, in which Z-plasty principles are used to lengthen the palate. Each of these techniques attempts to reconstruct the normal anatomy, including the nasal mucosal layer, the oral layer, and the muscle layer. To reconstruct the muscle sling, the abnormal insertions of the muscles from the back of the hard palate are divided, and the muscles are repositioned to re-create the levator sling across the midline. There is still debate in the literature regarding transaction of the tensor veli palatini tendon. When performed, it allows a more radical reposition and tension-free repair of the levator,^{135,136} although with possible detrimental effects on middle ear ventilation.¹³⁷ Flores et al¹³⁸ suggested tensor tenopexy as an alternative that improves Eustachian tube function and possibly reduces the need for future myringotomies.

Key Elements of Our Approach

Although the basic techniques used in dissection, intravelar muscle repair, and closure of the oral and nasal layers are repeated in each palate repair, the exact design of incisions, both medial and lateral, varies, depending on the anatomy and extent of each case and whether we are addressing a cleft of the primary and secondary palate (unilateral or bilateral) or a cleft of the secondary palate alone. We will discuss the common features, recognizing that the surgeon must vary the approach to fit the needs of the specific case.

The patient is taken into the operating room and a general anesthetic is administered. A Rye tube is used for endotracheal intubation, because it can be positioned out of the field of view for the repair. A prophylactic dose of cefazolin is administered before the procedure. When indicated, pressure equalization tubes are placed before the palate repair.

The patient is positioned at the end of the operating table with his or her head in a soft doughnut cushion. A roll is placed beneath the shoulders. The patient undergoes standard preparation with water-based chlorhexidine suspension and is appropriately draped.

The surgeon inserts a Dingman mouth gag and takes care to preserve the midline position of the endotracheal tube and not overly compress the tongue. The areas of projected incisions are injected with 0.5% lidocaine with 1:200,000 epinephrine. The same solution is then used to hydrodissect the mucoperiosteal flaps from the hard palate. After allowing 10 minutes for hemostasis, the surgeon incises the margins of the cleft. The tip of a No. 12 blade is inserted at the junction of the hard and soft palates and carried posteriorly to the uvula. The incision is then carried

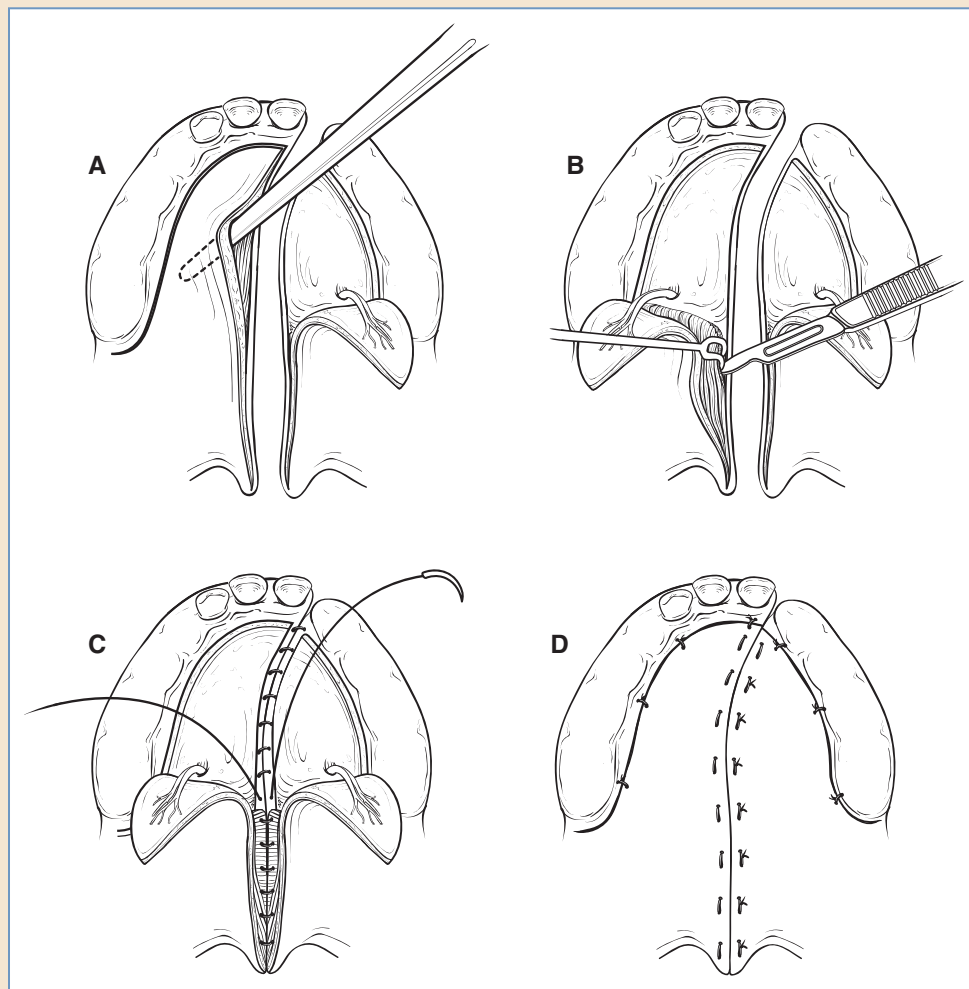


Fig. 22-7 Basic technique of the Salzer-Bardach two-flap repair. **A**, The oral and nasal mucosa are incised and separated from the uvula forward toward the incisive foramen on each side of the cleft. A second incision is completed on each palatal shelf to allow flap elevation. The second set of incisions goes from the anterior extent of the previous incision, continuing posteriorly at the junction of the palatal mucosa and alveolar mucosa. Relaxing incisions are made behind each maxillary tuberosity. **B**, With each full-thickness flap elevated and the greater palatine vessels preserved, the soft palate musculature is taken off its abnormal attachments to the posterior palatine bones. **C**, The nasal mucosa is sutured for a watertight closure from the incisive foramen toward the uvula. The soft palate musculature is sutured across the midline. **D**, The oral mucosa is closed from the uvula forward toward the incisive foramen.

anteriorly along the entire margin of the cleft, leaving a small cuff of nasal mucosa to facilitate nasal lining repair (Fig. 22-7). This incision is carried higher on the oral side of the hard palate in wider clefts to both gain additional nasal lining and have a sturdier border of tissue to suture.

With the oral mucosa placed under tension by a small double-pronged skin hook, the surgeon dissects the muscle from the oral mucosa by repeated gentle strokes with the belly of a No. 15 blade. Care should be taken not to overdissect the muscle from the oral mucosa, because many of the nerve fibers to the levator enter the oral surface of this muscle. The surgeon then moves the skin hook to grab the levator, and with this muscle held under tension, the surgeon dissects the nasal mucosa from the levator by repeated strokes of a No. 15 blade. In our experience, this maneuver facilitates precise and atraumatic dissection of the muscle fibers from the mucosal layers (see Fig. 22-7).

The surgeon makes stab incisions approximately 0.5 cm posterior to the greater tuberosities of the alveolus on either side. Through the stab incision, the surgeon introduces a dental scaler lateral to the hamulus of the pterygoid plates and gently fractures the hamuli on both sides. The incision is brought up from the stab incision toward the greater tuberosity, deviating toward the midline so the attached gingiva is not incised. The surgeon then brings the incision forward all the way to meet with the median incision of the cleft margin in a complete CL/P. For incomplete clefts, the surgeon extends the incision to a point approximately 1 to 2 cm in front of the anterior extent of the cleft (Fig. 22-8).

The dissection of the mucoperiosteal flaps off the hard palate begins from lateral to medial and is then extended posteriorly. The surgeon should identify and carefully preserve the greater palatine vessels that enter the flaps at the back of the hard palate.¹³⁹ With the flap retracted posteriorly, the aberrant insertion of the muscle to the posterior hard palate can be easily visualized and the muscle dissected free. The surgeon should accomplish this liberation both medial and lateral to the vascular pedicle to allow tension-free advancement of the flaps toward the midline. The surgeon dissects the nasal mucosa from the nasal surface of the palatine bones bilaterally with the use of a dental scaler or a gentle muscle hook.

In wide bilateral clefts, a vomer flap can be used to assist in the closure of the nasal layer. After the surgeon injects the vomer liberally with local anesthetic with epinephrine, the flaps should be incised in the midline, elevated bilaterally, and then reflected laterally to allow repair to the cuff of the nasal mucosa.

Careful hemostasis is achieved at this point. It is not unusual to encounter arterial bleeding from small branches of the greater palatine artery at the edges or undersurface of the flaps. This can be stopped with short strokes of bipolar cautery. Oozing from the areas of hamuli fracture is controlled by packing with small pieces of Surgicel gauze. The surgeon should repair the nasal mucosa first. The repair begins anteriorly and proceeds posteriorly to the back of the palate. We use interrupted 5-0 Vicryl sutures on a PS-4C reverse-cutting (hook-shaped) needle, with the knot facing the nasal cavity. The muscle layer is repaired with 5-0 clear nylon simple interrupted sutures with the knot on the nasal side. The surgeon typically places five or six such sutures throughout the length of the muscle from its most anterior border to the uvula. The oral mucosa is repaired with horizontal or vertical mattress 4-0 Vicryl sutures.

The keys to successful closure are well-vascularized flaps and a tension-free closure. Alteration of the position of the initial incisions to provide a sturdier edge of the nasal flap for nasal lining, adequate mobilization of the flaps based on the vascular pedicle only, and the change in the orientation of the flaps from vertical to horizontal are factors that bring the flaps closer together and decrease tension. The mattress sutures on the oral side make for a watertight closure with good surface approximation.

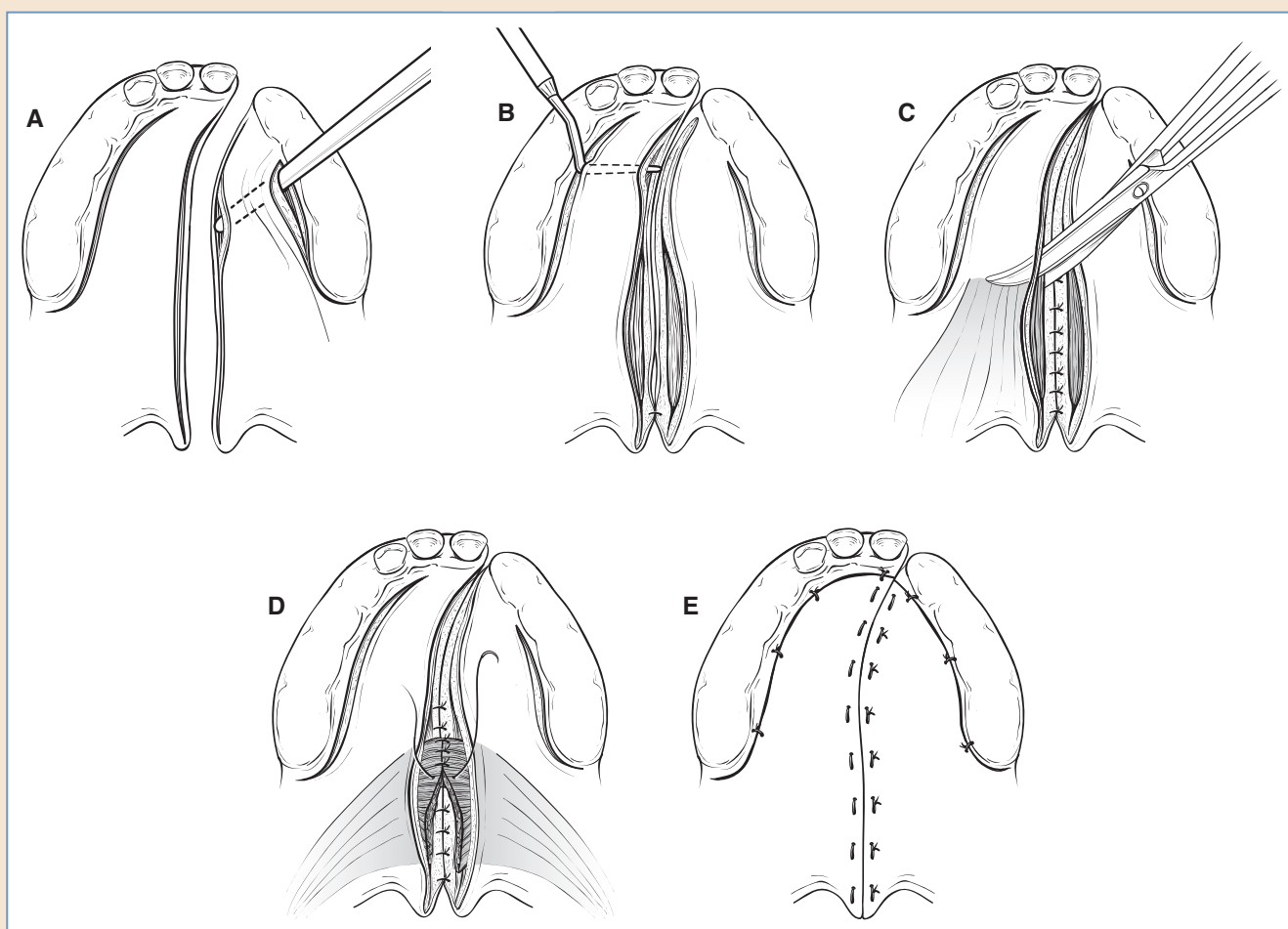


Fig. 22-8 Basic technique of the von Langenbeck cleft palate repair. **A** and **B**, Incisions and initial elevation of full-thickness mucoperiosteal palatal flaps are carried out anteriorly; the muscle dissection separates the oral and nasal mucosa in the soft palate. **C**, The abnormal levator muscle attachments are freed from the posterior hard palate. **D**, The nasal mucosa is repaired from just posterior to the alveolus to the uvula, and levator muscle reconstruction is completed. **E**, The oral mucosa is closed with interrupted sutures from the uvula to the posterior border of the alveolus. Laterally, the palatal flaps are left to heal by secondary intention.

Furrow's Double-Opposing Z-plasty

The double-opposing Z-plasty introduced by Furrow^{114,140} is an important alternative to the extended intravelar veloplasty described previously. We routinely use this technique for the primary repair of submucous cleft palates, and we also recognize its value as a secondary technique to lengthen the palate and correct velopharyngeal dysfunction when preoperative assessment indicates that the previous muscle was inadequate or that palatal lengthening alone may be sufficient to correct velopharyngeal dysfunction. Many centers successfully use this technique with slight modifications for primary palatal repairs.

The surgeon designs opposing Z-plasties on the oral and nasal mucosal surfaces (Fig. 22-9). The posteriorly based flap on each surface is composed of muscle and mucosa, and the anterior surface is composed of mucosa only. In most cases, the limbs of the Z form a 60-degree angle, but in shorter palates the angles are more obtuse. When the lengths of the two sides of the soft

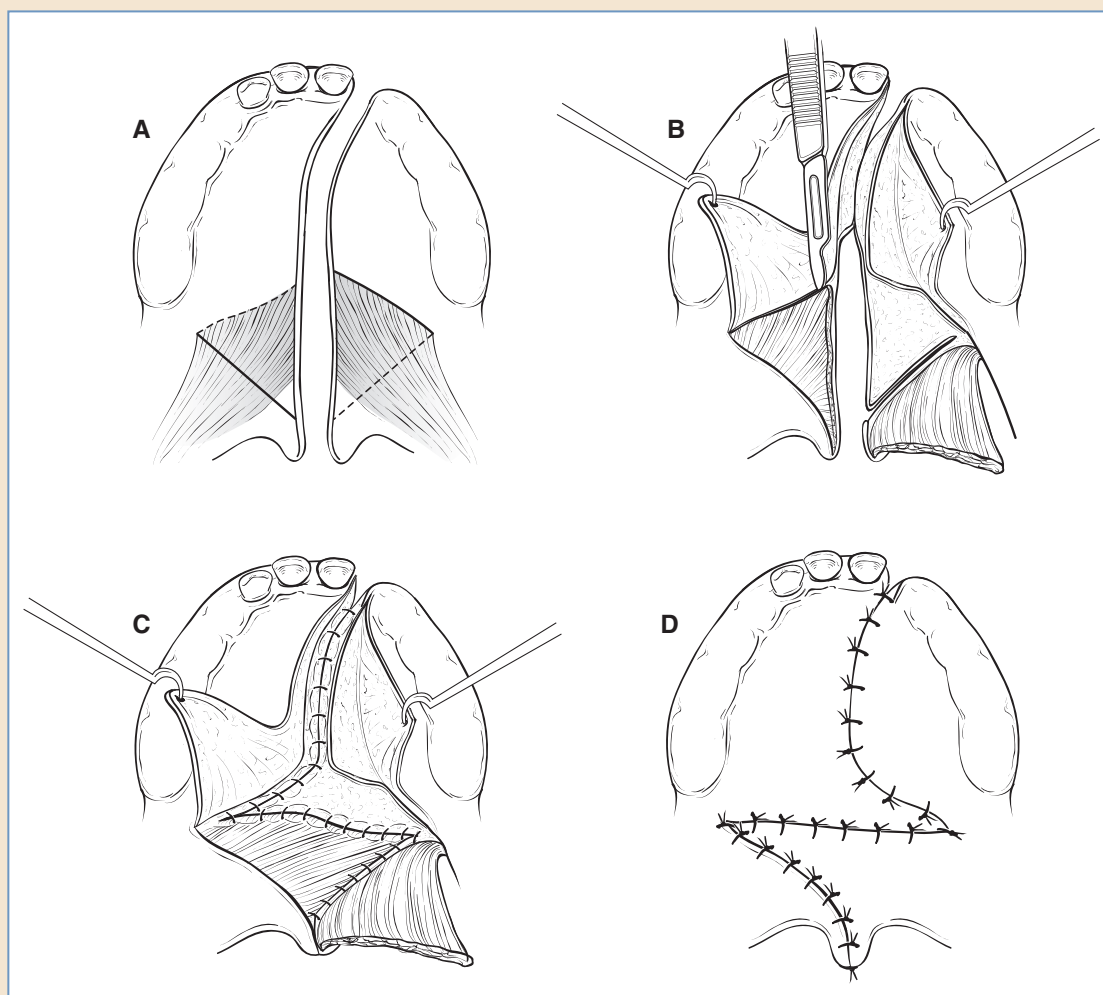


Fig. 22-9 Basic technique of the Furlow double-opposing Z-plasty palate repair. **A**, Location of planned incisions is indicated. **B**, Flaps are elevated. **C**, Nasal side closure is achieved with opposing Z-plasty flaps in the soft palate and nasal flap closure over the hard palate. **D**, Oral side closure of the hard palate and soft palate is completed.

palate are asymmetrical, asymmetrical angles are used. The left-sided limb of the Z is drawn from the position of the hamulus posterolaterally to near the junction of the soft and hard palate anteromedially. The right-sided limb is drawn from a point near the base of the uvula to a point near the hamulus.¹⁴¹

Although claimed unnecessary by Furlow himself, many surgeons use von Langenbeck-type lateral relaxing incisions, deepened into the space of Ernst, and infracture of the hamulus to allow additional medial movement of the palatal shelves. It seems that this extensive lateral release has found its way into the practice of many of the surgeons who use double-opposing Z-plasties for primary cleft palate repair.¹⁴⁰

Despite differences in surgical techniques, a general postoperative routine exists. A tongue stitch of 2-0 silk is placed at the end of the procedure and taped to the child's cheek. This stitch is normally removed the next morning. Hydration is maintained during the immediate postoperative

period with intravenous fluid. Oximetry is continuously monitored for 24 hours. Arm splints are also applied to prevent the child from disrupting the wound by placing fingers or sharp objects in the mouth. Oral feeding may be initiated postoperatively in the evening with a syringe or by having the patient drink from a cup. Nipple feeding is avoided for 10 days. Patients can usually be discharged with arm splints on postoperative day 1.

Treatment of Submucosal Cleft Palate

Submucosal cleft palate is usually characterized by the triad of bifid uvula, a bony notch at the back edge of the hard palate, and a translucent midline zone.¹⁴² In surveys of classic stigmata of submucosal cleft palate among the general population, the incidence has been reported to be 0.02% to 0.08%,¹⁴³ but the true incidence of submucosal cleft palate remains unknown, because many patients remain asymptomatic throughout life. Some patients presented with velopharyngeal inadequacy (VPI) during speech development. Others presented with hearing loss or recurrent ear effusions. The incidence of occult and classic submucosal cleft palate in patients with VPI is 10% to 22% and 17% to 44%, respectively.^{144,145} Many surgical approaches have been described for the treatment of submucosal cleft palate, including a pharyngeal flap, palatal pushback techniques, intravelar veloplasty, or a combination of these. A recent retrospective study by Ha et al¹⁴⁶ found that surgical correction is successful regardless of technique and age, but earlier recognition is advantageous for minimizing speech impairments. We think that the primary surgery for a submucosal cleft palate should be intravelar veloplasty, because the realignment of the velar musculature provides the greatest improvement in speech. This is achieved without the complications of the pharyngeal flap, such as sleep apnea and airway compromise.

Use of Facial Artery Myomucosal Flap for Palate Reconstruction

The *facial artery myomucosal* (FAMM) flap was originally conceptualized by Pribaz et al,¹⁴⁷ who combined the principles of nasolabial and buccal mucosal flaps. The flap has been used successfully in a variety of oronasal mucosal defects. The original series consisted of 15 patients, 4 of whom were patients with congenital cleft palates. The flap is a facial artery–based axial flap, which consists of mucosa, submucosa, a small amount of buccinator muscle, the deep plane of the orbicularis oris muscle, and the facial artery and venous plexus. The anatomic relationship between the facial artery and vein within the pedicle was further refined in a later article.¹⁴⁸ It can be designed either as a superiorly based (retrograde flow) or inferiorly based (anterograde flow) flap. It can also be used as a unilateral or bilateral flap. Relevant indications are oronasal fistula closure or palatal lengthening for VPI. Palatal lengthening with the bilateral FAMM should also be considered as the primary approach to palatal lengthening in patients in whom the palate is congenitally short, where there is no evidence of submucous clefting, or other anatomic anomaly other than shortness. The FAMM in this case allows the addition of tissue without the disruption of the intact musculature and does so as in other patients without a compromised airway (Fig. 22-10).

Surgical Technique: Facial Artery Myomucosal Flap for Palate Reconstruction

The course of the facial artery is outlined intraorally with Doppler ultrasonography. The flap is centered over the artery with an oblique orientation, extending from the retromolar trigone to the level of the ipsilateral gingival labial sulcus at the level of the alar margin (see Fig. 22-10). The width of the flap is 1.5 to 2 cm and is well anterior to Stensen's duct. Dissection starts at the distal end of the flap, with an incision through the mucosa and buccinator muscle. The facial artery is then identified, ligated, and cut. The rest of the flap is then incised, and the dissection proceeds just deep to the facial vessels, taking only a minimum of overlying buccinator muscle and orbicularis muscle near the oral commissure. A long pedicle (up to 8 to 9 cm in adults) can be raised,

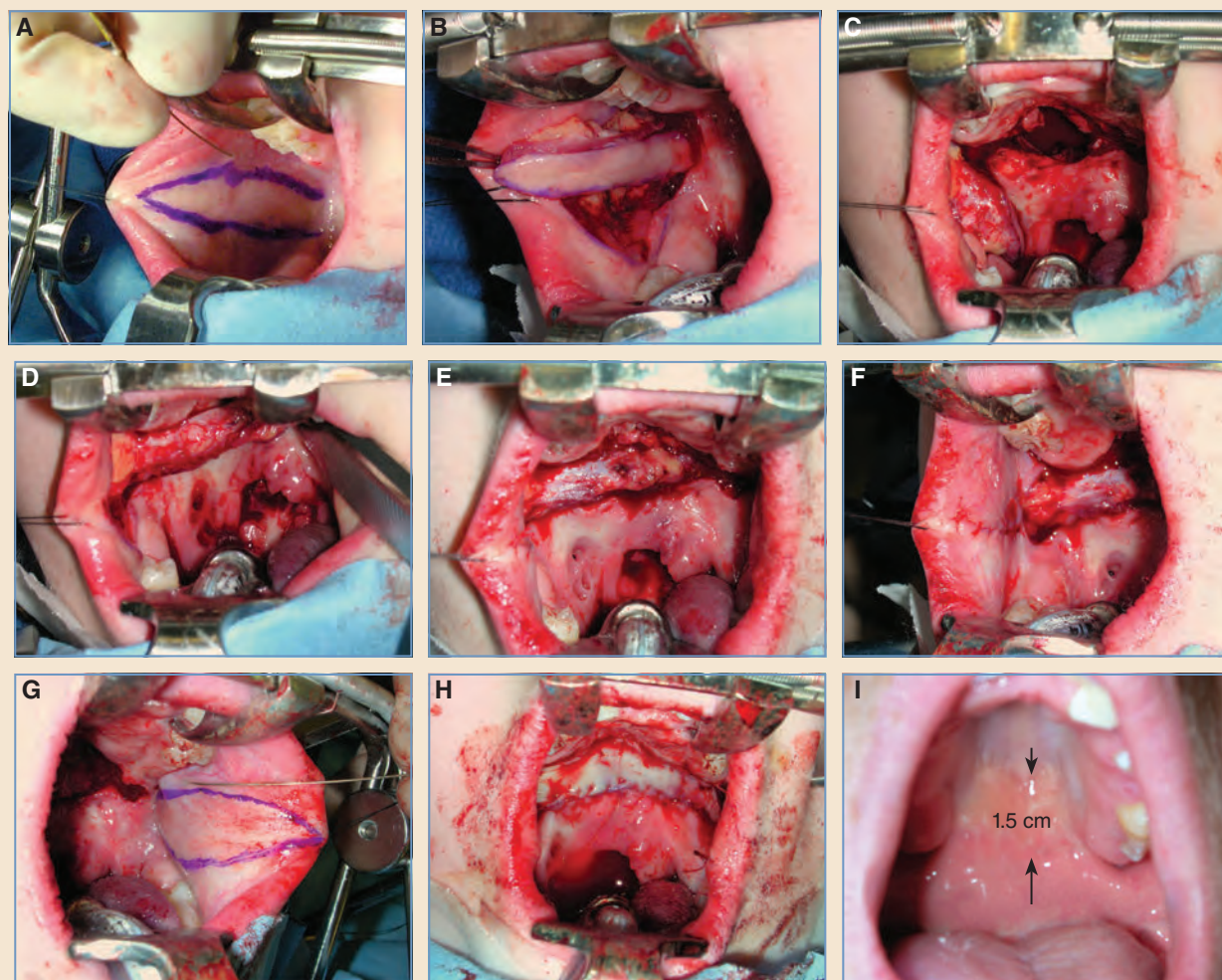


Fig. 22-10 **A**, With a probe in Stensen's duct, the 10 to 12 mm wide flap is oriented starting posterior to the maxillary tuberosity and tapered toward the oral commissure. Drawing and elevation facilitated by a traction suture at the oral commissure. **B**, The right-sided myomucosal flap elevated. **C**, The soft palate is released from the posterior hard palate with a full-thickness incision beginning a few millimeters behind the hard palate and extending on either side to the border of each flap. **D** and **E**, The right flap is rotated clockwise, turned over, and inset into the defect with the mucosal surface directed nasally. **F**, The donor site is closed in the right cheek. **G**, The left-sided flap is outlined. **H**, The left-sided myomucosal flap is rotated counterclockwise and inset on top of the right flap. The immediate lengthening effect on the soft palate is shown. **I**, Two years after surgery, the flap is well healed. The increase in length was 1.5 cm.

based either superiorly with a pivot point at the gingival sulcus or inferiorly with the pivot at the retromolar trigone. The donor site is closed, and care is taken to not to damage Stensen's duct. When the procedure is performed to close an oronasal fistula, the edges of the fistula are trimmed first. The FAMM flap is inset into the oral side of the fistula and sutured to the oral mucosa with 5-0 and 4-0 Vicryl sutures. When the procedure is performed for palatal lengthening, a long incision is made through all the layers of the palate at the junction between the hard and soft palates. One of the FAMM flaps is inset to reconstruct the nasal side of the oronasal fistula, whereas the contralateral flap is inset to reconstruct the oral side. The surgeon may find that it is easier to elevate each flap and close the greater part of each donor site, then make the transverse inci-

sion across the palate, and finally inset each flap as an alternative to the sequence shown in Fig. 22-10. In this technique the soft palate can be lengthened and pushed back toward the posterior pharyngeal wall. This approach is similar to the buccopharyngeal flap used by Hens and others,¹⁴⁹ emphasizing the desire to not disturb the nasopharyngeal diameter to preserve airway.

MANAGEMENT OF COMPLICATIONS

Complications related to cleft palate repair are generally infrequent. Complications include bleeding, airway compromise, infection, dehiscence, and fistula formation.¹⁵⁰ Significant bleeding is rare, but it may require a return to the operating room for exploration and hemostasis. In a retrospective analysis of 709 patients, there was a 2.4% rate (17 of 709) of take-back to the operating room in the immediate postoperative period for control of bleeding, although no blood transfusions were required.¹⁵¹ Airway compromise is also rare, but it may be life-threatening when present. Infants with Pierre Robin sequence or other congenital anomalies affecting the airway, either directly or indirectly, are at highest risk for airway problems.¹⁵²

Fistula formation is surgeon dependent and to a lesser extent depends on the technique used and the $\rightarrow$ type of cleft.^{150,153} The incidence of fistula formation has been reported to be between 0% and 34%, with the average between 3% and 4%. Most fistulas are small and are easily repaired during subsequent surgical procedures. Anterior fistulas near the alveolus are not repaired, because they are rarely symptomatic and are usually addressed at the time of bone grafting.^{154,155} Large fistulas or fistulas occurring in scarred, reoperated palates are difficult to repair. It is a common mistake to try repairing these defects with small local flaps—a strategy that almost always results in fistula recurrence, with further scarring of local tissues and subsequent limitation of reconstructive options. The surgeon should always use large flaps to successfully treat these defects. Large mucoperiosteal flaps, tongue flaps, FAMM flaps, and free microvascular tissue transfer should be considered in addressing this difficult reconstructive problem¹⁵⁶; the latter procedure is more commonly used to repair large fistulas after tumor extirpation.

VPI will be discussed in Chapter 23. Nonetheless, it is worth commenting here that the issue of airway compromise is one of the major concerns that is faced in the decision of treatment of nasal air emission (NAE). The approaches range from repeat repair with intravelar veloplasty if not done, Furlow technique if the gap is small and the palate is short, to some type of pharyngoplasty. We think that a greater degree of palatal lengthening can be obtained with the use of the bilateral FAMM (as described previously) or the buccopharyngeal flap. Both techniques add significantly more length than the Furlow Z-plasty and avoid airway compromise.

In a large multicenter study,¹⁵⁷ secondary surgery after isolated palate repair was done in 13.6% of patients by the age of 16 years, with higher rates in complete clefts. The risks of secondary surgery over time showed a bimodal distribution; the first peak within the first postoperative year and the second peak between 3 and 5 years after surgery. The first risk peak after secondary surgery reflects the short-term complications that must be addressed within the first postoperative year. The second peak reflects the longer-term complications diagnosed at the age at which children reach speech milestones.

OUTCOMES

The most important outcomes in cleft palate repair are the establishment of normal speech and hearing.¹⁵⁸ Confirmation of proper muscle repositioning and velar function can be assessed with

MRI and videofluoroscopy imaging, respectively. These issues, however, do not necessarily correlate with speech impairment.¹⁵⁹ The most important step for achieving good outcomes is a well-executed surgical repair by an experienced surgeon. Once palate repair has been performed and pressure equalization tubes have been placed, frequent evaluation by a multidisciplinary cleft team is necessary so that adjunctive measures can be undertaken when speech abnormalities are identified. Many patients with cleft palate have some language delays and articulation errors; therefore early speech intervention is both recommended and beneficial.

Despite a well-executed palatoplasty, careful speech follow-up, and early intervention, about 15% to 20% of patients will develop speech production disorders, most commonly velopharyngeal dysfunction. This percentage seems independent of the type of palatoplasty.¹⁶⁰ Velopharyngeal dysfunction is diagnosed clinically by a constellation of symptoms that include pathologically incurred nasal resonance (hypernasality), misarticulation, escape of air through the nose (nasal emission), and aberrant facial movements (grimacing).¹⁶¹⁻¹⁶³

Successful outcome analysis in cleft surgery is difficult to obtain for a multitude of reasons.¹⁶⁴⁻¹⁶⁹ Much of the cleft literature is descriptive and focuses on anatomic integrity and aesthetic results.^{170,171} More recently, comparative studies have been reported that assess functional outcome with respect to speech,¹⁷² dentoskeletal development,¹⁷³ psychological issues, and airway.¹⁷⁴ Critical information on long-term outcomes in cleft care is hard to come by. The time lapse between surgery and measurement of outcome (at craniofacial growth maturity in mid to late adolescence) creates major difficulties for clinical researchers.^{170,171,175} Retrospective study requires information on primary management practices 5, 10, and 15 years previously and presupposes clearly described treatment protocols. Even when this exists, such practices may have little relevance to the research questions of current interest. Where treatments have been described with insufficient detail, treatment-related variation and outcome may remain undetectable. Similarly, if the results of prospective studies are to be timely and relevant, it is important that they be completed within a reasonable length of time. Unfortunately, the measurements of some aspects of outcome (for example, dentoskeletal development) are best deferred until patients reach maturity.

Comprehensive examination of the outcome of treatment should be multifaceted, especially because trade-offs may occur. For example, delayed closure of the hard palate may reduce growth disturbance but increase speech impairment. Evidence that adequate palatal closure has a salutary effect on the ubiquitous problem of middle ear pathology, the possible benefits of avoiding disuse atrophy in the levator veli palatini musculature, and the early establishment of swallowing and crying indicate that closure may provide many benefits if done much earlier. None of these factors has been clarified by objective studies. Prospective, longitudinal investigations of the clinical management of CL/P are necessary to answer the three central problems of that management: the best time to treat, the techniques of treatment most appropriate in a particular patient, and the effects of treatment.^{163,176} The treating physician must keep in mind all of the goals listed previously to provide an environment that promotes a successful outcome.

CONCLUSION

The management of patients with cleft palate is complex. The functional goals of cleft palate surgery are to facilitate normal speech and hearing without significantly affecting facial growth of the child. To achieve these goals, cleft palate teams must provide multidisciplinary care and monitor the functional outcomes of surgery.

KEY POINTS

- Oral clefts are of multifactorial cause, with both genetic predisposition and environmental influences playing a role.
- Orofacial clefting is genetically complex, with no single gene responsible for all forms.
- Clefting can result from a single gene defect either as part of a syndrome (for example, Van der Woude syndrome, Treacher Collins syndrome, and velocardiofacial syndrome) or as an isolated phenotypic effect (for example, X-linked cleft palate and nonsyndromic, autosomal dominant orofacial clefting).
- Potential environmental risk factors that have been associated with oral clefts include smoking, alcohol, caffeine, benzodiazepines, and corticosteroids.
- With the higher incidence of syndromal association with cleft palate alone, all patients should be evaluated by a geneticist.
- Given the potential association of Stickler syndrome with Pierre Robin sequence, all babies with Pierre Robin sequence should have a screening by a pediatric ophthalmologist.
- It is important to accurately analyze and report a cleft lip and/or palate deformity in a standardized manner. Kernahan's classification scheme (modified by Millard and others) is reported on a diagrammatic Y-shaped symbol, with the incisive foramen represented at the focal point.
- The premaxilla, alveolus, and lip, which are anterior to the incisive foramen, are parts of the primary palate. Structures posterior to it, which include the paired maxilla, palatine bones, and pterygoid plates, are part of the secondary palate.
- The pathologic sequelae of cleft palate include feeding and nutritional difficulties, recurrent ear infections, hearing loss, abnormal speech development, and facial growth distortion.
- The muscle sling of the levator veli palatini is interrupted by cleft palate. In patients with cleft palate, the effectiveness of the velar pull against the posterior pharyngeal wall is impaired, and velopharyngeal incompetence results.
- The abnormality in the tensor veli palatini increases the incidence of middle ear effusion and middle ear infection in children with cleft palate. Early evaluation by a pediatric otolaryngologist is imperative.
- An intact velopharyngeal mechanism is essential for production of nonnasal sounds and is a modulator of airflow in the production of other phonemes that require nasal coupling. Speech impairments specific to velopharyngeal dysfunction include sibilant consonants, the sounds *s*, *z*, *sh*, *zh*, and *ch*; *j* or soft *g*; and the intraoral pressure consonants *p*, *b*, *t*, *d*, and *k* and hard *g*.

- Cleft palate maxilla has some intrinsic deficiency of growth potential. Growth potential is further impaired by surgical repair. The growth disturbance is exhibited most prominently in the prognathic appearance during the second decade of life despite the normal appearance in early childhood.
- Palatoplasty is performed at 6 to 12 months of age, depending on the extent of the cleft. Patients with Pierre Robin sequence and those with idiopathic micrognathia typically undergo repair at 12 to 14 months of age unless the airway is severely compromised.
- In Furlow's palatoplasty, the posteriorly based flap on each surface is composed of muscle and mucosa, and the anterior surface is composed of mucosa only.
- In intravelar veloplasty, the muscle layer is repaired with 5-0 clear nylon simple interrupted sutures with the knot on the nasal side. Five or six such sutures are typically placed throughout the length of the muscle from its most anterior border toward the uvula.
- The FAMM flap is a reliable surgical modality for persistent oronasal fistula after cleft palate repair.
- Submucosal cleft palate, often undiagnosed until speech development, may lead to VPI, chronic middle ear effusion, and hearing loss.

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23

The Surgeon's Role in Management of Velopharyngeal Dysfunction

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Circular Closure Pattern of the Velopharyngeal Mechanism; Coronal Closure Pattern of the Velopharyngeal Mechanism; Submucous Cleft Palate With Sphincter Closure Pattern of the Velopharyngeal Mechanism.



elopharyngeal dysfunction (VPD) is a distinct malfunction of the velum and pharyngeal musculature that can cause a collection of speech and swallowing disorders. The causes can be multifactorial, but the fundamental problem is anomalous velopharyngeal closure that prevents normal speech production. The reasons for this may be anatomic, neurologic, behavioral, or a combination of these. Some have structural abnormality (such as a cleft palate) and others have a neurologic problem (for example, myasthenia gravis). In this chapter we will focus on the structural abnormalities. Most patients who have untreated VPD will develop compensatory maladaptive articulations in an effort to be understood by others. Patients often present with hypernasality, nasal emissions, or facial grimacing during speech. Failure of the sphincteric mechanism is frequently the result of a structural defect of the pharyngeal walls or soft palate at the level of the nasopharynx.

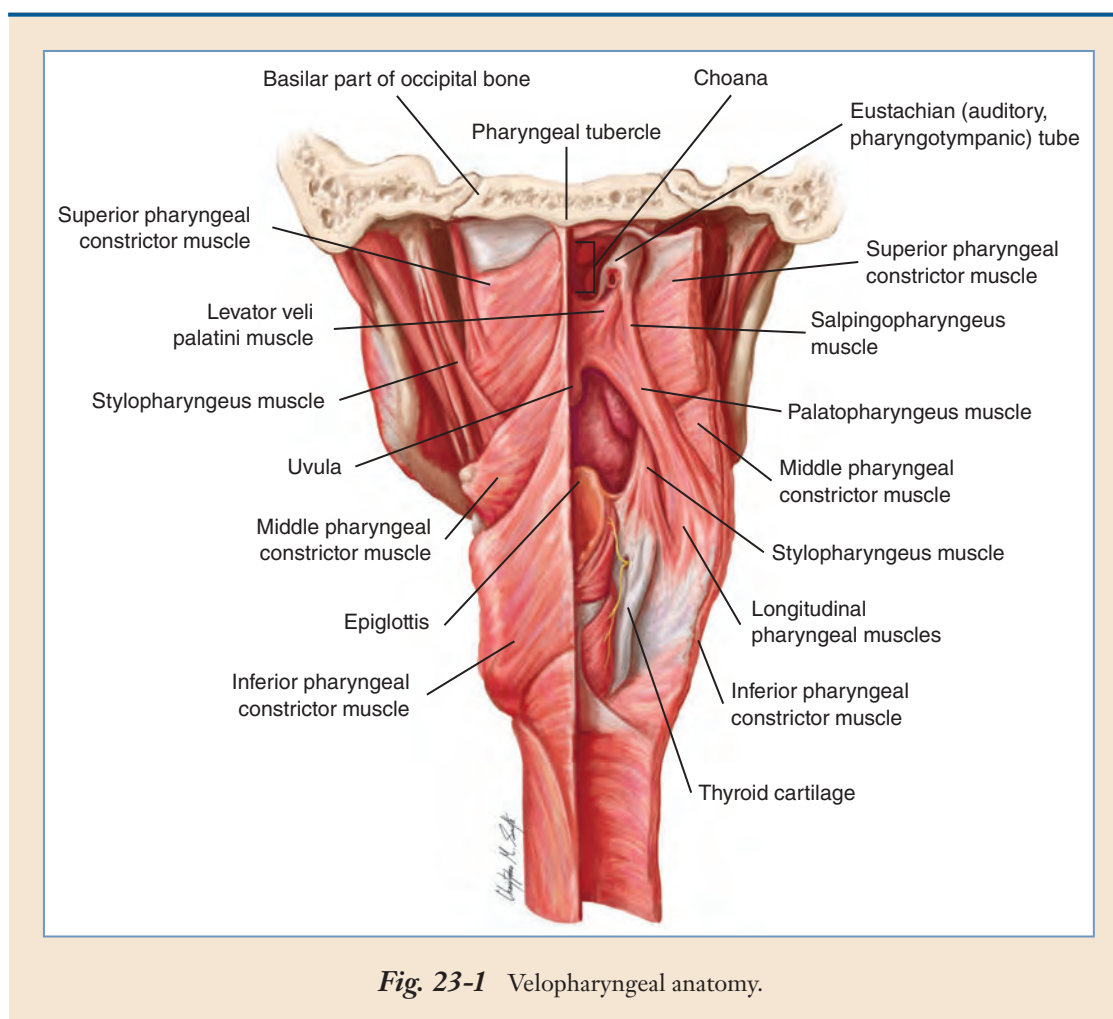
This chapter discusses the causes of VPD, the approach to diagnosis and management, and surgical approaches for optimizing velopharyngeal sphincter function while preventing airway morbidity.

ANATOMY

Velopharyngeal Mechanism

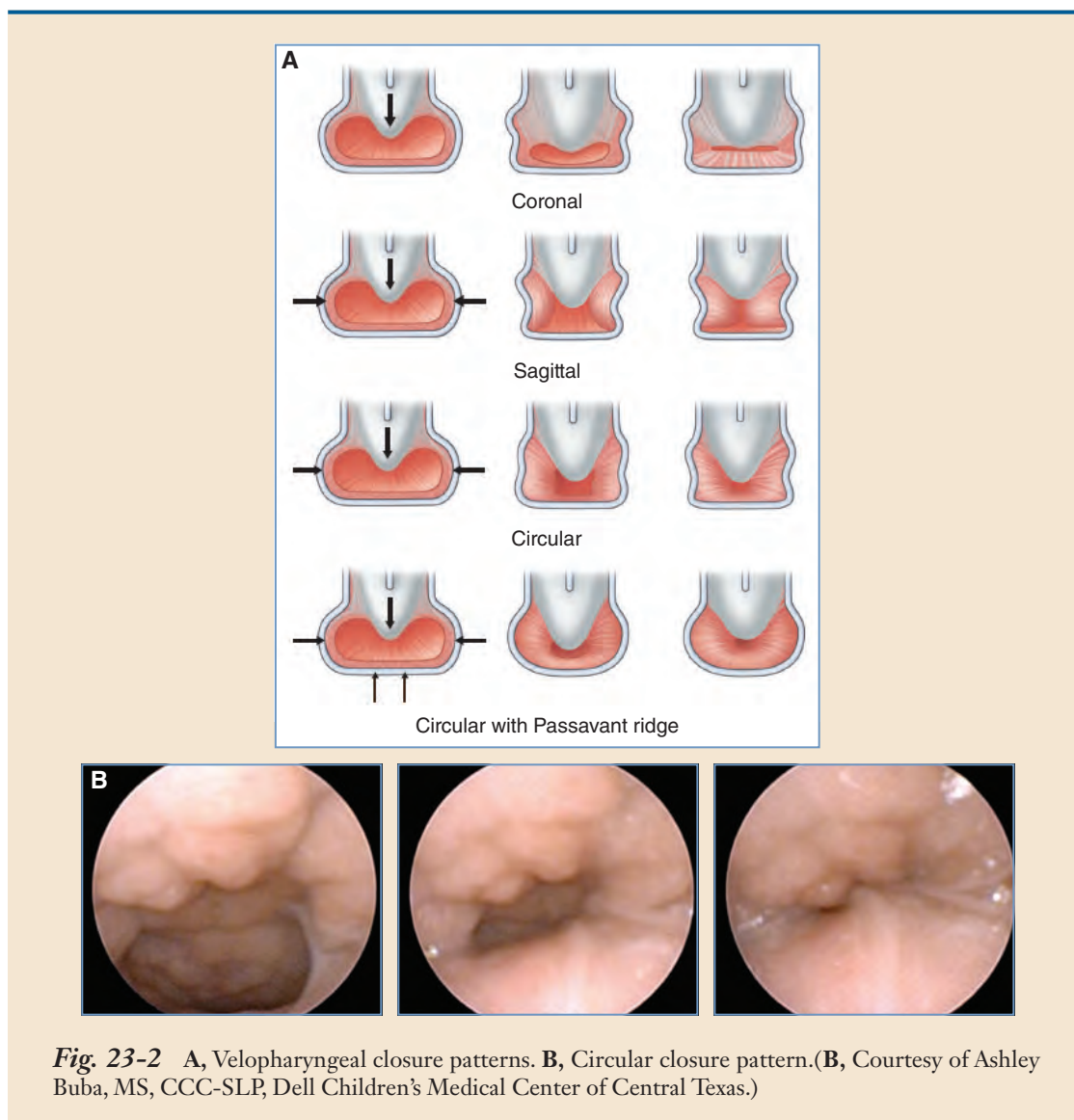
Within the region of the velopharynx, multiple muscles work together in normal circumstances to close what is called the *velopharyngeal sphincter*. The two anatomic structures involved are the velum, or soft palate, and the pharynx, including the lateral and posterior pharyngeal walls. When functioning in concert, the muscles of these structures close off the nasopharynx, a critical movement for normal speech production and swallowing. The principle muscles that constitute the velopharyngeal sphincter include the levator veli palatini, the palatopharyngeus, and the superior pharyngeal constrictor. Lesser contributions come from the tensor veli palatini, the palatoglossus, the musculus uvulae, and the salpingopharyngeus¹ (Fig. 23-1).

The tensor veli palatini is innervated by cranial nerve V (trigeminal nerve); the rest of the muscles are innervated by contributions from cranial nerve IX (glossopharyngeal nerve) and cranial nerve X (vagus nerve) where they form the pharyngeal plexus.

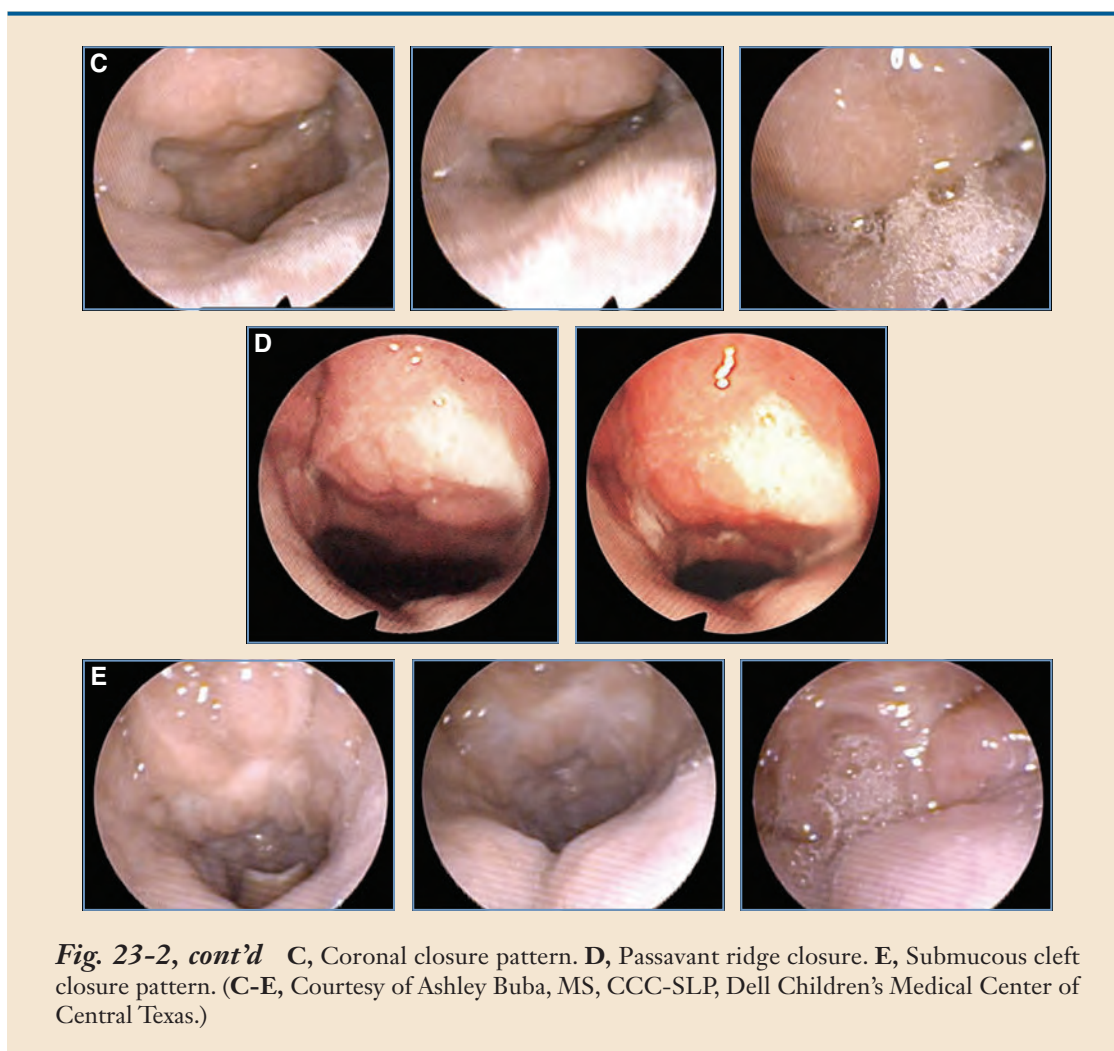


Closure Patterns

The composite movements of the lateral and posterior pharyngeal walls and the velum close the velopharyngeal port in deglutition and during oral speech sounds. These movements open the port for breathing and some nasalized articulations. Despite the variance in how the velopharyngeal mechanism achieves closure, characteristic patterns of movement have been observed using direct visualization with nasopharyngoscopy. These typical patterns of closure include coronal, sagittal, circular $\pm$ the Passavant ridge, and bow-tie-type closure¹ (Fig. 23-2, *A* through *E*).



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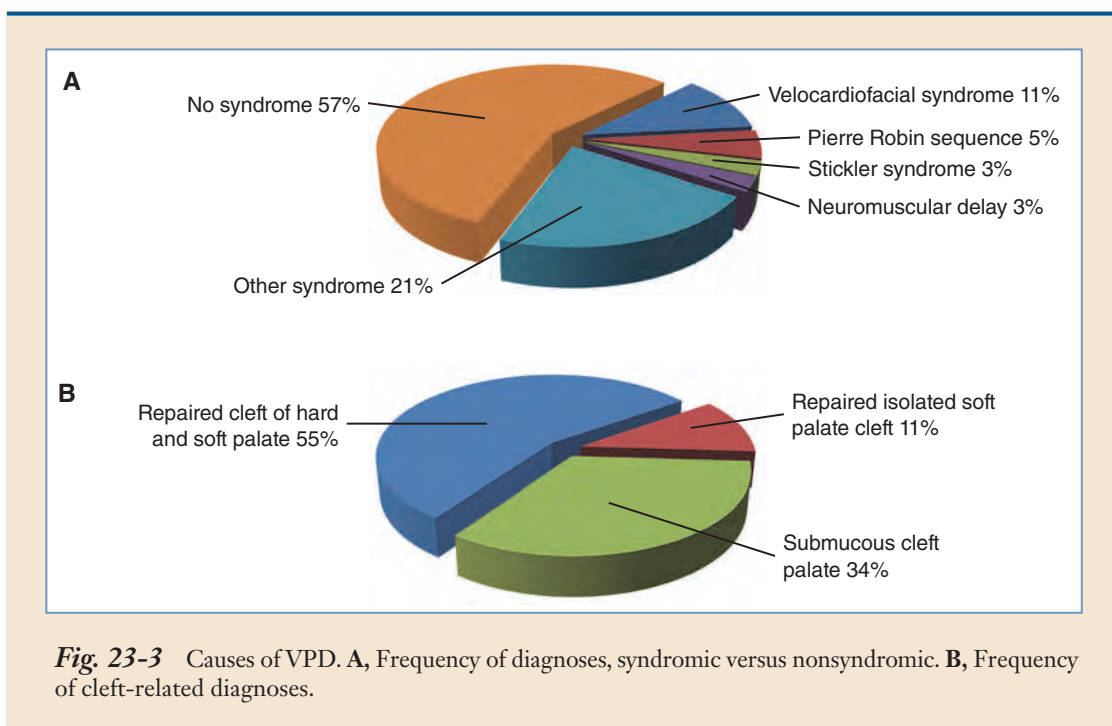


ETIOLOGIC FACTORS

In this chapter we will discuss the concept of VPD that is present as speech develops. In a study of 148 patients, some causes of secondary VPD have been established² (Fig. 23-3). This is distinguished from speech problems resulting from primary cleft palates in that it has numerous causes, not all related to cleft palate (Box 23-1). For information on cleft palate, please refer to Chapter 22.

Secondary Cleft Palate Dysfunction

The normal anatomy of the velopharyngeal mechanism is altered in cases of cleft palate. In addition to the concomitant disruption of the mechanism by the cleft space, muscular function is altered, especially that of the levator veli palatini, a major contributor to velar excursion during speech. During proper primary repair of the cleft palate, the levator muscles should be liberated from the abnormal insertion on the hard palate.



Box 23-1 Common Causes of Velopharyngeal Dysfunction

- Secondary cleft palate dysfunction
- Submucous cleft palate
- Velocardiofacial syndrome (22q)
- Tonsillectomy
- Midface advancement

A subset of patients presents after cleft palate repair with VPD. Whether from improper surgical technique or uncontrolled contracture, these patients have a scarred, immobile, and often short soft palate, inadequate to maintain appropriate closure of the velopharynx. A post-operative oronasal fistula may also cause VPD. Its presence and contribution to hypernasality are established and considered in the overall treatment plan. However, these fistulas do not contribute to VPD.

Submucous Cleft Palate

Submucous clefts can be difficult to identify. They occur where mucosal continuity over a levator muscle is discontinuous and oriented as it would be in a cleft palate. Calnan described a triad of findings in submucous cleft palates, including the zona pellucida, a bifid uvula, and a palpable notch at the posterior hard palate. Speech findings in patients with submucous cleft palate can run the gamut from normal or near-normal to grossly abnormal^{3,4} (Fig. 23-4).



Fig. 23-4 Submucous cleft palate. (Courtesy of Robert J. Shprintzen, PhD, President, The Virtual Center for Velo-Cardio-Facial Syndrome, Inc.)

Velocardiofacial Syndrome (22q11.2)

22q11.2 deletion syndrome is known by many names, including *DiGeorge syndrome*, *velocardiofacial syndrome* (VCF), *Shprintzen syndrome*, and *CATCH22*, among others. The abundance of names is a side effect of the variability of the syndrome. It affects approximately 1 in 4000 people. Inheritance is considered autosomal dominant; however, most cases are not inherited. Del 22q has many possible signs and symptoms that can vary widely, even among people affected within the same family. The findings in del 22q commonly include conotruncal cardiac malformations at a rate as high as 74%. Palatal abnormalities may be present in up to 69% of patients and include VPD, submucous cleft palate, bifid uvula, and cleft palate. A host of other associated abnormalities can occur that are beyond the scope of this text.^{5,6}

Bezuhly et al⁷ reported that their series of 23 patients with 22q and submucous cleft palates repaired with Furlow Z-plasty or pharyngeal flaps had similar results between groups. In the 22q group, they noted the need for more revisions and an increased median time to normal resonance by a factor of 5. In their study, these patients were 3.6 times less likely to achieve normal resonance.

Anomalous internal carotid arteries have been shown to be a frequent feature of velocardiofacial syndrome. These vessels pose a potential risk for iatrogenic injury and hemorrhage during velopharyngeal narrowing procedures. Iatrogenic injuries to the carotid artery during velopharyngeal surgery are strikingly absent in the literature. Occasionally, transmission of vascular pulsations through floppy, redundant mucosa may be mistaken as an ominous vessel. Careful preoperative evaluation and dissection will frequently obviate the need for imaging studies. Our center does not order routine angiography of these patients before speech surgery. Baek et al⁸ showed that such imaging is not cost effective (Fig. 23-5).

Tonsillectomy

Although not common, a tonsillectomy and an adenoidectomy may lead to VPD. There are case reports of cranial nerve injuries in the vicinity of the operative field leading to postoperative VPD. Additionally, removal of bulk from the pharynx may contribute mechanically to VPD, preventing complete closure presumably from the prior adaptation of the existing musculature to the

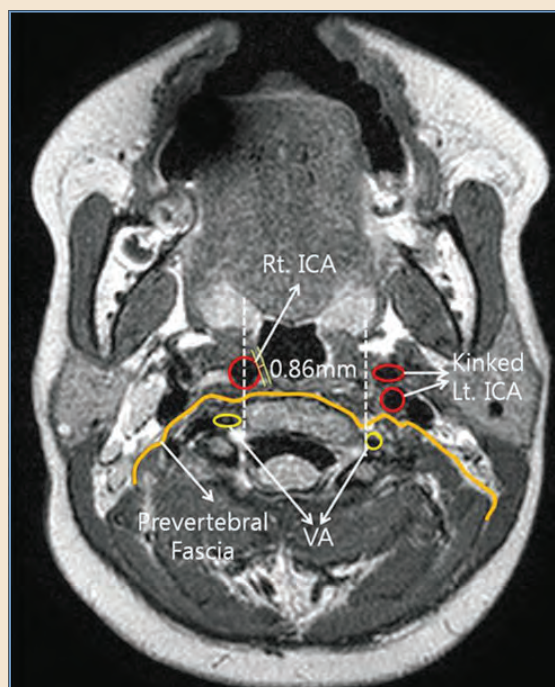


Fig. 23-5 MRI showing a medially displaced right internal carotid artery (*Rt. ICA*). (*A*, Artery; *Lt. ICA*, left internal carotid artery; *V*, vein.)

large, space-occupying tonsils and adenoids. A number of posttonsillectomy patients with VPD had an undiagnosed submucous cleft palate. Other identified causes of VPD in posttonsillectomy patients are neuromuscular disorders, poor palate mobility, behavioral disorder, postoperative nasopharyngitis, and scarring.⁹

Midface Advancement

Midface advancement with LeFort-level osteotomies is a common procedure for managing malocclusion and/or orbitomalar correction in cases of orofacial clefting and craniofacial syndromes. There are several predictors for the development of VPD. A short palate and a deep pharynx on cephalometric evaluation has been associated with postoperative VPD after a LeFort I-level osteotomy in cleft patients.¹⁰ Another clear predictor of VPD after midface advancement is the total distance of advancement. Longer distances are associated with a deeper pharynx and a higher chance for VPD. Finally, patients with preoperative VPD are more likely to have increased VPD after a midface advancement. Initially after midface correction, VPD may be present. A formal evaluation of speech should not begin until approximately 6 months after these procedures, because VP adaptation can occur, lessening the need for a formal evaluation and surgical treatment. In cleft patients with severe maxillary hypoplasia, midface distraction osteogenesis has been shown to have improved results over traditional orthognathic surgery, especially for distances of more than 10 mm.^{11,12}

EVALUATION

Coordination of Care

Patients with VPD should be managed by an interdisciplinary team. Critical members of the team addressing speech should include a craniofacial surgeon, a skilled speech-language pathologist (SLP), and a geneticist. A team approach is essential for customized treatment planning and better outcomes.¹³

Patient History and Physical Examination

When taking a history and performing a physical examination for patients presenting with VPD, surgeons should first focus on determining the underlying cause. (These were discussed previously.) Each of these is approached as a unique problem. In general, team members determine whether a patient's speech is intelligible and whether it has caused psychosocial problems or frustration, because others, including parents, siblings, peers, and teachers have difficulty understanding it.

During a physical examination, the size of the adenoid pad and the tonsils and the presence of fistulas are noted. A palpable palatal notch at the junction of the soft and hard palate, a zona pellucida along the soft palate, and a bifid uvula may be clues that a patient has a submucous cleft palate.

Another important detail to note is a history of obstructive sleep apnea (OSA). If this a concern preoperatively, then polysomnography may be indicated before speech surgery, because the condition can worsen.

Speech Assessment

The hypernasality caused by VPD results from abnormal resonance in the vocal tract or abnormal airflow. The symptoms of VPD are auditory-perceptual; therefore the standard for assessment is a trained listener's judgment. Most SLPs associated with a cleft palate team report that they rely primarily on listener's judgment of resonance and articulation in evaluating velopharyngeal function.¹⁴ This is a specialty area in speech pathology, and not all SLPs have the skills to judge the characteristics associated with VPD.¹⁵ The role of SLPs is to help determine whether a patient has VPD and whether it is caused by structure, neuromotor deficits, or abnormal speech learning.

Perceptual Testing

An SLP will complete formalized articulation testing and will have the patient engage in syllable repetition and sentence repetition. A spontaneous connected speech sample is obtained. Therapists use appropriate speech samples to obtain the information needed for a definitive diagnosis.¹⁶ Speech samples need to be developmentally appropriate but also contain the appropriate types of phonemes.

Sentence repetition is a good way to evaluate articulation and velopharyngeal function. Using a battery of sentences such as those in Table 23-1, clinicians can quickly assess articulation, nasal emission, and resonance in a connected speech activity.¹⁶ This is a quicker and more valid assessment of normal speech production than a single-word, formal articulation test.^{16,17} Articulation and resonance are assessed during connected speech, which increases the demands on the velopharyngeal valving system to maintain closure. Hypernasality and nasal air emission are more apparent in connected speech than in single words or sentences. An increase in articulation errors is common during the production of longer, spontaneous utterances.¹⁵

Table 23-1 Sample Sentences for Assessment of Articulation and Resonance

p	Papa plays with a puppy.	f	I have five fingers.
b	Buy baby a bib.	v	He visits the cave.
m	Mama makes lemonade.	l	I like yellow lollipops.
w	Wade likes the water.	s	Sissy sees the sky.
y	You have a yellow yo-yo.	z	Zip up the zipper.
h	He has a big horse.	sh	She goes to the shop.
t	Take Teddy a toy.	ch	Chad rides the choo choo.
d	Daddy does dishes.	j	John told a joke to Jim.
n	Nancy is not here.	r	Ross had a red fire truck.
k	I like cookies and crackers.	er	The teacher is a doctor.
g	Go get the wagon.	th	That's my toothbrush.
ng	Put the ring on the finger.	blends	Splash, sprinkle, street

Visual Detection

A mirror examination helps to determine whether nasal air escapes while a patient produces oral sounds. A mirror is held under the nares during the production of pressure sensitive sounds. Nasal air emission is noted as condensation (fogging of the mirror). Consistent fogging on the mirror across all sound productions suggests decreased velopharyngeal closure from structure or neuromotor causes. Inconsistent fogging on the mirror can indicate abnormal speech learning, especially if it is isolated to one sound class (for example, sibilants).

Nasal grimacing is another visual indication of nasal air escape. It presents as a contraction near the nasal bridge and/or on the side of the nose. It occurs as an overflow muscular response to excessive effort in producing velopharyngeal closure.¹⁶

Instrumental Assessment

An experienced SLP can assess velopharyngeal function during speech production. However, if speech characteristics associated with VPD are noted, instrumental assessment can help to identify the source and nature of the deficit or dysfunction.¹⁸ Several types of instrumentation are available for evaluating VPD.

Direct Assessment

Direct instrumental assessment allows an examiner to observe the components of the velopharyngeal mechanism at rest and during functional activities. Frequently reported direct assessment techniques include multiview videofluoroscopy, nasendoscopy, and MRI.¹⁹ Nasendoscopy and videofluorography are the preferred instruments for direct visualization of the velopharyngeal mechanism during speech production.²⁰ Many cleft palate–craniofacial teams in the United States have decided that radiation is no longer needed for evaluating velopharyngeal function and have chosen nasendoscopy as their primary assessment tool.²¹ We will focus on nasendoscopy and videofluoroscopy.

Nasendoscopy and Video Fluoroscopy

Nasendoscopy provides a view of the velopharyngeal mechanism from above, showing the velum, posterior pharyngeal wall, and lateral pharyngeal walls during speech. The adenoid tissue and tonsils, which can either assist or interfere with closure, may be visible. Proponents of nasendoscopy report that it provides extensive information about velopharyngeal closure patterns and the contributions of the velum and pharyngeal walls.²² It has also been shown to be the most appropriate instrumental measure for diagnosing an occult submucous cleft palate and for evaluating velopharyngeal closure after a pharyngeal flap surgery.²³ Reported limitations of nasendoscopy include difficulty identifying small velopharyngeal gaps, difficulty viewing both lateral pharyngeal walls simultaneously, poor visualization because of glare from the light source, interference from adenoid tissue, and obliquity of view.^{13,24,25} Nasendoscopy can be performed by any well-trained professional on the team, but usually the surgeon or the SLP conduct this procedure.

Videofluoroscopy allows examiners to evaluate velopharyngeal movement and timing. Barium is given through the nose, which facilitates visualization of the margins of the velopharyngeal structures. The patient then repeats the target speech sample. Lateral images are obtained to assess velar movement posteriorly and superiorly. This view also shows posterior pharyngeal wall movement. Frontal images are obtained to view lateral pharyngeal wall movement. Because neither view can provide complete information regarding velopharyngeal closure, multiple views should be used to assess the depth of the nasopharynx, velopharyngeal movement, coordination, and the gap size.¹³ Limitations include the need to coordinate with a radiologist, radiographic exposure, and some controversy related to the types and number of images needed for an accurate assessment.²⁵

Regardless of the direct assessment tool employed, images should be measured and reported in a standardized manner to determine the most effective treatment.¹³ Golding-Kushner et al¹³ recommended the use of a ratio scale that estimates the velopharyngeal gap size compared with the size of the velopharyngeal valve at rest. This rating scale is used for both nasendoscopy and videofluoroscopy (Fig. 23-6).

Indirect Assessment

Indirect measures do not facilitate direct visualization of the velopharyngeal structures; they provide information about the functional outcomes of these structures. This allows the examiner to make inferences about the velopharyngeal competence. Common indirect measures include nasometry and aerodynamics assessment,¹⁹ which can be useful in monitoring patients' progress. For example, preoperative and postoperative data can be used to assess improvement.

Nasometry

Nasometry provides a ratio of sound energy transmitted through the nose and mouth during speech. It is an estimate of the average percentage of acoustic energy transmitted through the nose and through the mouth during speech. Nasalance scores are obtained while the speaker repeats words or sentences that, when spoken by normal speakers, contain little nasal resonance. Nasal resonance is expected to be minimal during these productions. Nasalance studies that surpass a predetermined ratio are sometimes used as evidence of VPD. Nasalance scores are most useful in tracking patients with VPD over time and in determining the effectiveness of an intervention at altering oral or nasal resonance for speech.²⁶

Aerodynamic Assessment

Aerodynamic evaluation is useful for assessing nasal airflow and oral air pressure to quantify the effects of VPD on the aerodynamics of speech. A mean oral air pressure of less than 3 cm H₂O and nasal airflow in excess of 300 ml/sec during the production of oral pressure consonants are

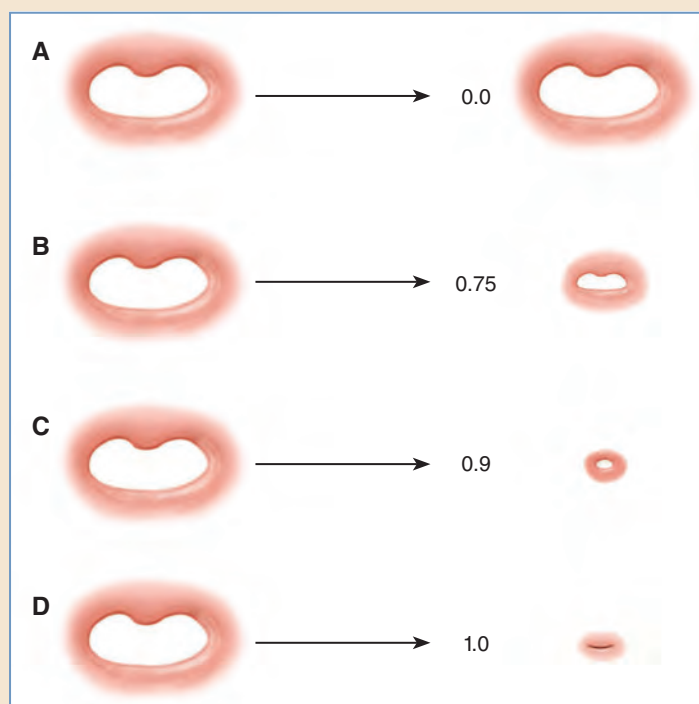


Fig. 23-6 The ratio of the velopharyngeal gap size at rest and during speech. The gap size is estimated, compared with the size of the velopharyngeal valve at rest. The gap may be estimated or calculated using computer programs that compute surface area.

considered in the abnormal range. A larger VP opening is usually indicated by lower oral pressure measurements and greater nasal flow measurements. From these measurements, inferences can be made regarding abnormal VP opening during production of pressure consonants; however, the reason it occurs cannot always be determined, especially in a patient with incorrect speech learning.²⁷

Imaging

Various forms of cervical vascular imaging studies, such as CT, MRI (angiogram), and traditional angiography, have been advocated as aids to surgery, because they define the preoperative vascular anatomy. Nevertheless, it is not known whether these studies alter either the performance or outcome of operations on the velopharynx. Many surgeons opt to perform these procedures without any preoperative imaging with good outcomes. Surgeons should be aware of the possibility of anomalous carotid arteries. Visual inspection and palpation intraoperatively guide cautious surgeons to perform safe speech surgery.

NONSURGICAL TREATMENT OPTIONS

Speech Therapy

Speech therapy is indicated to eliminate VPD caused by abnormal speech learning. Compensatory misarticulations in a patient with the ability to achieve velopharyngeal closure, phoneme-specific hypernasality, and phoneme-specific nasal air emission should be treated with speech therapy.



Fig. 23-7 A speech obturator with hard and soft palate components.

Speech therapy can be attempted in patients with borderline or inconsistent velopharyngeal closure. If imaging shows that a patient achieves near-complete velopharyngeal closure, speech therapy may help to improve velopharyngeal closure for speech. Therapy may focus on improving weak or imprecise articulation and improving oral airflow.²⁷ Kuehn et al²⁸ documented some success using continuous positive airway pressure during speech production to treat hypernasality.

Appliances

Prosthetic management may be a solution for treating VPD in a small number of cases. A palatal lift is an option for patients with adequate tissue but poor control of coordination and timing of velopharyngeal movements. A speech bulb, or obturator, is an acrylic mass used for closing velopharyngeal gaps (Fig. 23-7). This is an option in patients without adequate tissue. Prostheses can also be used to provide diagnostic information in patients with variable VPD. If it is unclear whether surgery would provide significant improvement in speech quality, a prosthesis may be used as a temporary tool to help guide surgical decision-making.

SURGICAL TREATMENT OPTIONS AND TECHNIQUES

Surgical Treatment Plan: Algorithmic Approach

Once prospective surgical candidates have undergone nasopharyngoscopy, the results are interpreted by a speech surgeon and SLP. The patient's underlying diagnosis will be placed into the context of the visualized velopharyngeal function to determine a treatment option (Figs. 23-8 and 23-9). Our center uses three options for surgical treatment of VPD: a Furlow palatoplasty, a sphincter pharyngoplasty, and a pharyngeal flap (superiorly based). We tailor the surgery to the specific diagnosis and nasopharyngoscopy findings (Table 23-2). For patients with a cleft palate who underwent a prior straight-line repair and have a small to medium velopharyngeal gap, a Furlow lengthening is selected. The caveat is a significantly short immobile palate. A Furlow procedure in this case would not be sufficient to restore velopharyngeal function, and a pharyn-

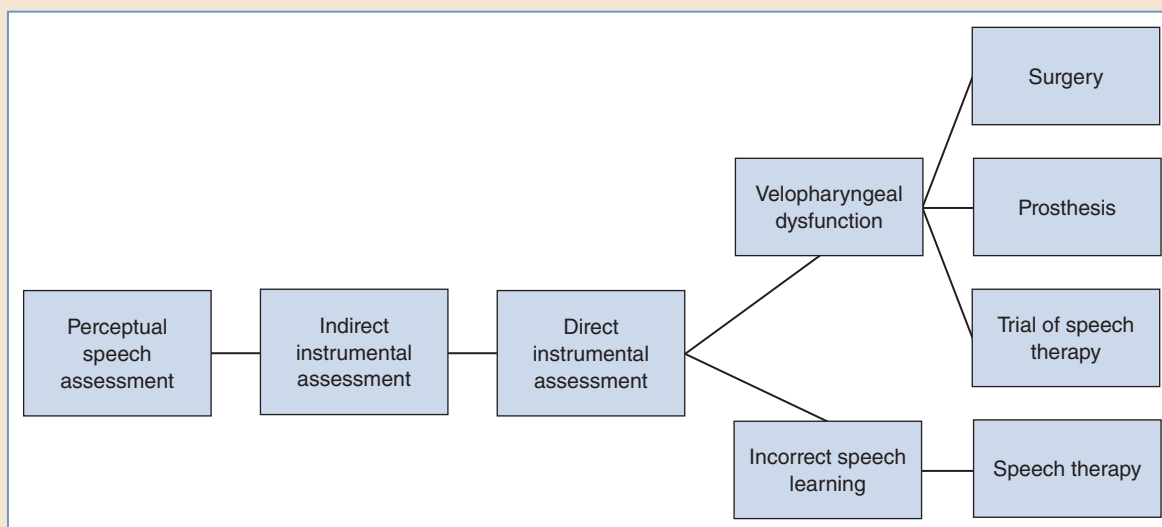


Fig. 23-8 The progression of assessment and treatment of VPD.

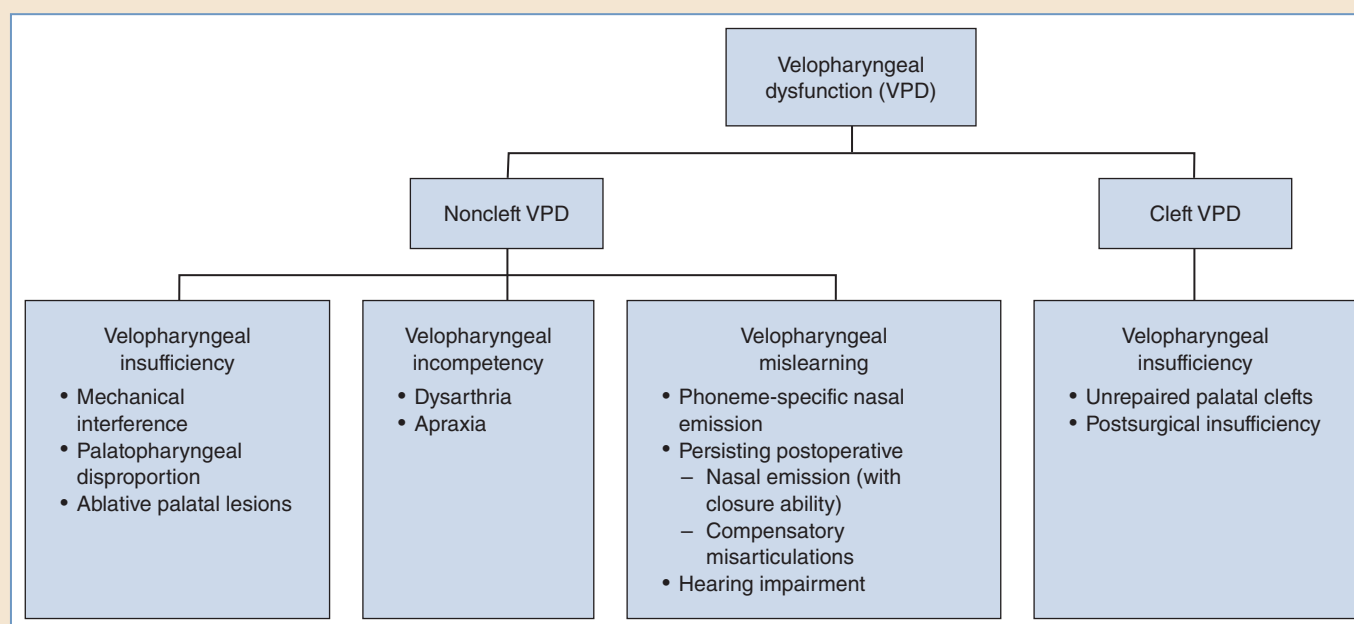


Fig. 23-9 Classification of VPD.

geal flap is chosen. In patients with reasonable palatal length and function but poor lateral wall movement, sphincter pharyngoplasty works well. Finally, medium to large velopharyngeal closure gaps, with either negligible or moderate lateral wall movement, are best treated with a pharyngeal flap. In general, we prefer a Furlow procedure and sphincter pharyngoplasty, whenever possible, over a pharyngeal flap because of the risk of OSA. Furlow palatoplasty and sphincter pharyngoplasty are more physiologic. We have found that following an algorithmic approach to surgical

Table 23-2 Tailoring Velopharyngeal Dysfunction Surgery to the Diagnosis and Scope Findings

	Furlow Palatoplasty	Sphincter Pharyngoplasty	Pharyngeal Flap
Diagnosis	Cleft palate with straight-line repair Submucous cleft palate (nonsyndromic)	Cleft palate, VCF, after adenotonsillectomy, after midface advancement	Cleft palate, VCF, after adenotonsillectomy, after midface advancement
Nasopharyngoscopy	Moderate gap size Palate excursion/movement	Good palate excursion Limited lateral wall movement	Large AP gap Lateral wall motion

selection is advantageous, even in patients with VCF, because of the variety of velopharyngeal closure patterns, and because not all patients have decreased muscle tone.²⁹

Management of the Palatine Tonsils

Many patients who require a surgical procedure to treat VPD have generous-sized tonsils. The approach for managing the tonsils in speech surgery patients differs from that in other patients with standard indications for removal. Although the tonsils may contribute to velopharyngeal closure, removing them would actually increase VPD in the short run. Planned removal approximately 2 months before VPD correction allows increased precision in designing and executing the preferred speech operation.³⁰ This is especially advantageous when a sphincter pharyngoplasty and a pharyngeal flap are indicated. On occasion, tonsils can be large enough to obscure key anatomic landmarks necessary for a successful operation.

Furlow Palatoplasty (Modified)

The various incisions, tissue planes, and limited access associated with performing a Furlow palatoplasty can create confusion for novice surgeons. A consistent approach using the proper instruments and assistance greatly facilitates efficient and successful surgical execution, preventing unnecessary struggle and cervical muscle strain for a surgeon and assistants.

The setup is as important as any other part of the procedure. A microcuffed right-angle endotracheal tube fits properly within the tongue blade groove of a Dingman mouth gag and prevents air leaks that cause bubbling of blood in the oropharynx, interfering with proper visualization. The use of a local anesthetic with epinephrine helps in two ways. The epinephrine induces vasoconstriction in a highly vascular field, and the anesthetic reduces pain-associated elevation of blood pressure, reducing bleeding in the field. The mouth gag is positioned, and the palate is widely injected before the patient is prepared to ensure adequate time for epinephrine to take effect (approximately 10 minutes).

This description of the procedure and approach assumes a prior palatoplasty or a submucous cleft, where the presenting symptom is velopharyngeal insufficiency, usually in patients older than infancy. The procedure is also described for the development of an oral-side myomucosal flap on the left and a nasal-side myomucosal flap on the right.

The initial incisions for a Furlow palatoplasty are made within the hard palate at least a centimeter anterior to the hard palate–soft palate junction, with attempted preservation of a triangular extension of the hard palate. The apex is pointed posteriorly in the midline, providing an anchoring point for hard palate flaps and setup for a V-Y pushback of the hard palate. This maneuver serves to relieve tension on the suture lines in the soft palate. Starting within the hard palate eases the dissection and identification of the greater palatine vessels, whose preservation is important to successful healing because of their vascular supply to the region. Raising the hard palate tissues off the posterior palatine bone also facilitates visualization and suturing of the nasal-side flaps, which will promote success as well.

The incision is started at the lateral margin of the hard palate near the maxillary tuberosity and curved at the maxillary tuberosity laterally behind the dentition to prevent inadvertent injury to the vessels. The cut proceeds anteriorly and laterally and ends at a point about 1 to 2 cm anterior to the maxillary tuberosity. The incision is angled in an AP-medial direction to create a point in the middle of each hard palate flap, and then it is angled posteromedial to the midline, meeting the opposite side again about 1 to 2 cm anterior to the posterior edge of the hard palate margin. (The incision on the hard palate looks like a W.) Fig. 23-10 shows the W-shaped incision on the hard palate, which sets up the V-Y advancement of the hard palate flaps. Subperiosteal dissection should proceed posteriorly to identify the greater palatine vessels. If a midline scar is present, separating the oral mucosa from the nasal mucosa is helpful, leaving the nasal side intact. Preserving the tension associated with an intact nasal scar or uninterrupted mucosa facilitates dissection of the soft palate flaps. Once the hard palate flaps are raised, stay sutures are placed in the apex of the flaps, with a couple throws to assist with retraction.

Once the greater palatine vessels are identified emerging from their posterolateral foramen, dissection should continue around the vessel with a Cottle elevator, including dissection along the posterior edge of the palatal bone as it curves to join the medial aspect of the medial pterygoid plate. Caudal retraction on the hard palate flaps facilitates exposure. The lesser palatine vessel will be encountered lying in a horizontal plane at the junction of the palatine bone and medial pterygoid plate just posterior to the greater palatine vessel. It should be cauterized, because it will tether the flaps, causing undesired tension on transposition. Bleeding can be encountered here as small vessels that perforate the medial pterygoid plate are disrupted, but this will be self-limited. Freeing the soft palate from the medial pterygoid plate eases the dissection of the oral and nasal flaps. An incision is made with a No. 11 blade through all layers of the posterior three quarters of the soft palate in the midline.

With the surgeon standing at the head of the patient, the assistant places a single, fine hook deep to the oral mucosa, close to the start of the incision just made. The hook should not catch the nasal soft tissues. Lateroposterior retraction with a toe-in motion and slight elevation will help to reveal the palatal glands that start at the posterior edge of the palatine bone. The use of monopolar electrocautery on a low, blended setting with an angled Colorado tip can reduce bleeding, enhancing visualization of the targeted plane between the palatal glands and the palatal muscle complex whose fibers run at an angle. A broad dissection plane should be maintained to avoid working in a hole. Proceeding posteriorly, it becomes obvious that the plane comes closer to the oral mucosa as the thickness of the palatal glands diminishes. Once the nasal-side myomucosal flap is separated from the oral-side palatal gland–mucosal flap, an elevator is used to further dissect the tissues off the medial pterygoid in a subperiosteal plane. If a bony cleft is present at the posterior edge of the hard palate, a hockey-stick elevator can be used to strip the nasal floor some degree to release tension.

At this point, the assistant should move to the left side of the surgeon and place the hook in the opposite side with same type of toe-in motion. A Cottle elevator is used to release the tissues off the posterolateral edge of the hard palate toward the medial pterygoid plate to facilitate dis-

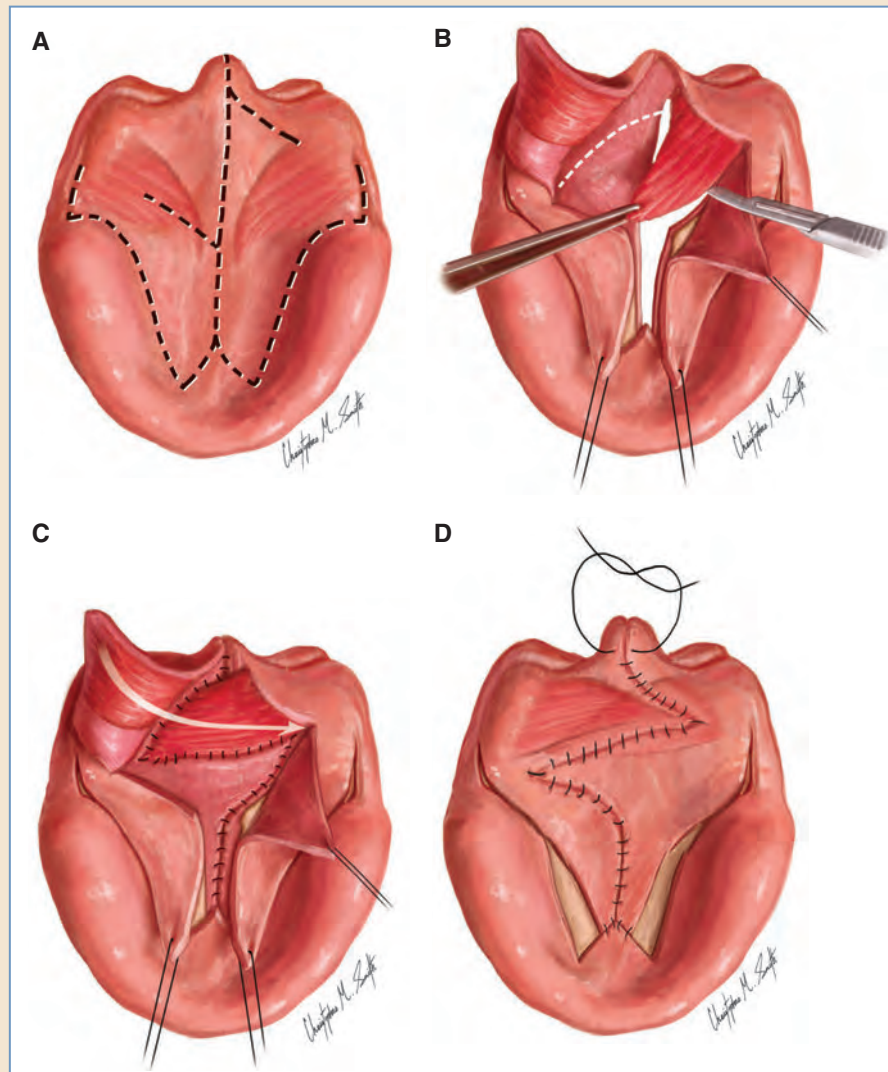


Fig. 23-10 W-type incision on the hard palate for a Furlow palatoplasty. **A**, Markings for a modified Furlow palatoplasty. **B**, Completed oral-side myomucosal dissection on the patient's left and oral-side palatal-gland mucosal flap on the right. The nasal-side myomucosal flap on the right is actively being developed. The nasal-side mucosal flap on the left is marked. **C**, Completed nasal-side reconstruction with interdigitating flaps. Note the reorientation of the muscle. **D**, Completed oral closure. Note that there is some raw surface area over the lateral hard palate; this heals secondarily.

section by opening up the planes of dissection. The dissection plane between the muscle complex and the nasal mucosa is the most difficult dissection of the procedure. Constant attention to the angle of the nasal mucosa helps to prevent inadvertent button holes in the thin mucosal flap. The use of face-lift scissors with modified tips, such as Kaye face-lift scissors, also helps with dissection. Bipolar electrocautery assists in stopping the small mucosal bleeders, which is necessary to maintain visualization of the dissection plane. Once posterior and lateral dissection is sufficient,

the nasal mucosa can be stripped off the medial pterygoid and nasal floor in the same fashion as on the right side using a mirror hockey-stick elevator.

Several maneuvers are helpful for mobilizing the tissues in preparation for transposition of the Z-plasty flaps. As mentioned previously, subperiosteal dissection of the nasal-side flaps off the medial pterygoid and nasal floor liberates these flaps quite effectively. If a lateral relaxing incision is used in the soft palate, the tensor veli palatini tendon and hamulus of the medial pterygoid plate will be revealed. Either infracture of the hamulus and/or lysis of the tendon will relieve tension. Additional subperiosteal dissection medial to the incision will finish the release of the soft palate. Last, performing an osteotomy of the posteromedial aspect of the greater palatine foramen with a 2 mm osteotome and liberating the vessel from the canal can relieve additional tension, if present. This maneuver typically provides an additional 2 to 3 mm of medial advancement per side. Experience with the procedure will aid surgical decision-making. Ultimately, patients missing large quantities of tissue, such as those with Pierre Robin sequence, will require more release maneuvers than patients who are born with less severe deformities.

Z-plasty flaps are designed and cut to rotate the muscle complex from their anomalous oblique course, with insertion onto the posterior edge of the hard palate to a transverse, overlapping position. When the dissection is carried out as described, with the right muscle complex attached to the nasal mucosa, the proximal cut is on the right for the nasal Z-plasty closure. Care is taken to make sure this cut is a couple millimeters posterior to the hard palate edge and roughly parallel to the anteriormost fibers of the muscle complex. Leaving a rim of nasal mucosa attached to the posterior edge of the hard palate eases placement of the sutures. Flaps are transposed and sutured while ensuring the mucosa is everted toward the skull base for submucosa-to-submucosa contact. Horizontal mattress sutures can help to achieve proper closure when needed. A small, curved needle (P-2, Ethicon) driven by a long Castroviejo needle holder can be used for placing sutures in this deeper position.

After closure of the nasal side, the oral-side Z-plasty flaps are fashioned with the most proximal cut on the left side to properly rotate the oral-side myomucosal flap to a transverse position. Because the oral-side Z-plasty is not as deep, it is easier to use a larger, thinner needle such as an RB-1 or TF. To kill dead space that naturally occurs as the flaps tent over the nasal closure, the oral closure near the posterior edge of the hard palate can take a bite of the nasal closure on the first pass of a vertical mattress suture. If tension is released off the stay sutures on the hard palate flaps, these flaps can be pushed back in V-Y fashion before the vertical mattress suture is placed. The vertical mattress suture helps to secure the V-Y advancement, relieving tension on the oral Z-plasty closure. The anterior portion of the hard palate flaps is secured to the triangular extension of the midline hard palate. The uvula can be repaired by resecting one of the two uvulas present or by excising the medial half of both and suturing the two halves. Exposed vessels or bone laterally can be covered by splitting the superior pharyngeal constrictor behind the maxillary tuberosity with blunt dissection and allowing the cheek fat pad to herniate into the field. Several sutures can be placed to position the fat that will be mucosalized by secondary intention.

Discussion

The procedure described previously is a modification of the original double-opposing Z-plasty described by Leonard Furlow.³¹ Most surgeons who perform this surgery today use lateral incisions in the hard palate and lateral relaxing incisions in the soft palate. This allows closure of a wider cleft using the Furlow technique. A cleft team working in an interdisciplinary fashion to treat secondary VPD can use nasopharyngoscopy to determine the anatomic derangement of the velopharyngeal mechanism. A subset of patients has a history of a straight-line cleft palate repair; their palates are sagittally deficient and show mild to moderate excursion, with a coronal gap. A Furlow palatoplasty can be performed to successfully treat both VPD and undiagnosed submucous cleft palate in this subgroup.³² Chen et al³³ studied the use of Furlow palatoplasty

for treating secondary VPD related to cleft palate and found it to be effective in 16 of 18 (89%) cases. Their criteria for the Furlow procedure in these patients were:

1. Marginal VPD regardless of age
2. A velopharyngeal closure gap of less than 5 mm
3. A short, scarred palate with an abnormal levator position
4. Good lateral wall movement with a short palate

In a series of 148 patients, a Seattle group² treating secondary VPD patients found a small velopharyngeal gap size to be the most important indicator of success of a Furlow palatoplasty.² In their cohort with a 57% nonsyndromic diagnosis of cleft palate repair (or submucous cleft palate), 73% had resolution of VPD when the gap size was small. As gap size increased, success diminished.

Sphincter Pharyngoplasty

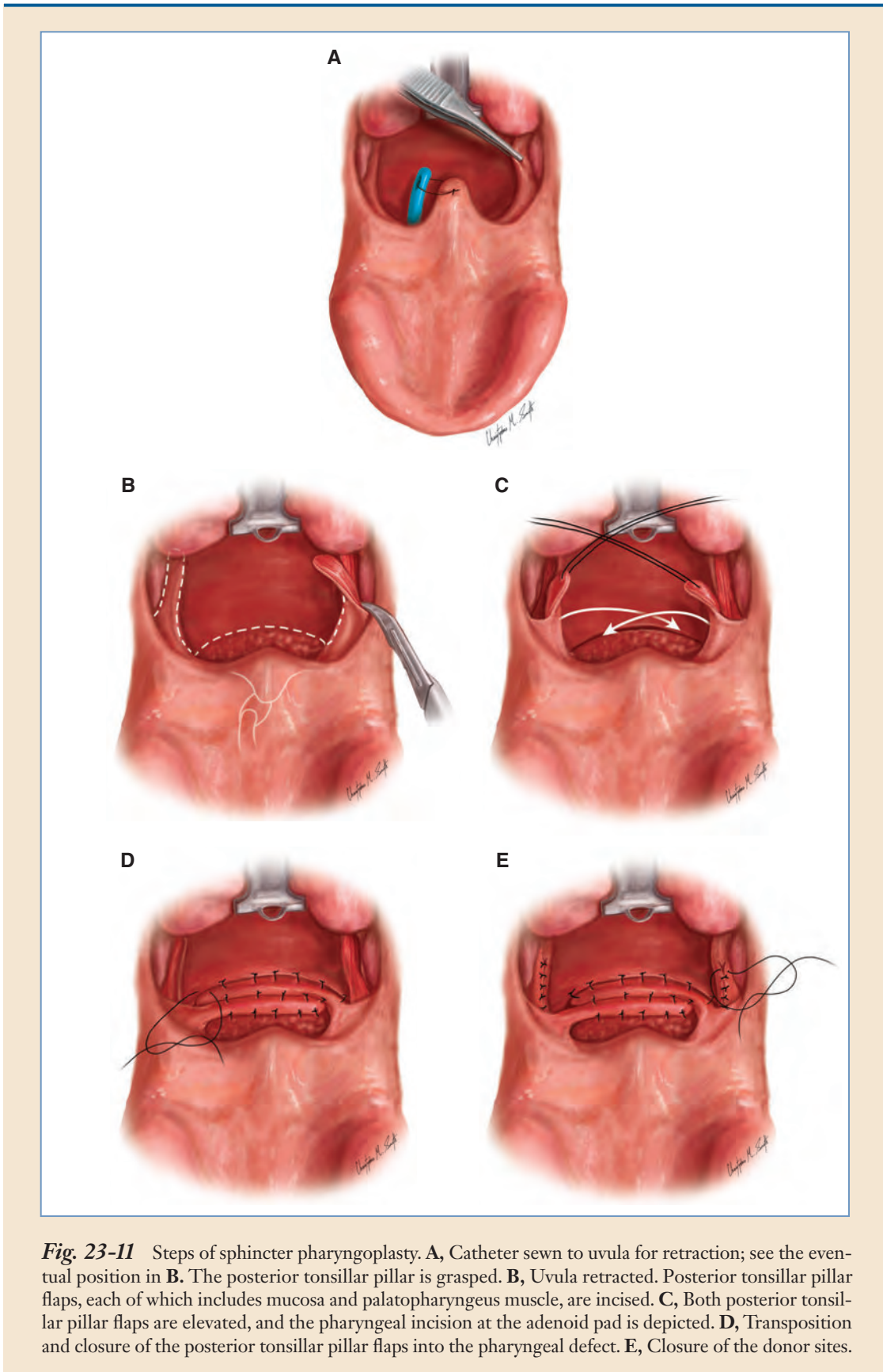
Positioning and Marking

The patient is placed in a supine position on the operating table. After induction of general anesthesia and control of the airway with an oral right-angle endotracheal tube taped in the midline, the table is turned for access to the head. A shoulder roll and gel roll for the head are placed. The neck is extended, and a Dingman mouth gag is inserted. The Dingman has several features of adjustability that can optimize exposure. First, an appropriate-sized blade is selected. This will allow adequate retraction of the tongue without the blade becoming an obstructive force, that is, too large a blade will obscure part of the posterior pharynx. For most cases (in school-age children), a No. 2 blade is appropriate. But this must be tailored to the patient's age and size. Second, when a Dingman is placed, it is inserted until the second row of tooth hooks is secure on the central incisors. Third, the tooth retractors are maintained together in the midline for maximal excursion of the Dingman. Once the Dingman is opened (preoperatively), lidocaine with epinephrine is injected into the posterior soft palate, the posterior tonsillar pillars, and the posterior pharynx, especially along the adenoid pad. The surgeon may attempt to mark the proposed posterior tonsillar pillar flap; however, with the redundancy in the posterior pharynx, this is often futile. The mouth gag is released, and the patient is prepared and draped.

Technique

The Dingman is reopened, with the tongue centered on the blade. If airway pressures increase (through communication with anesthesia), the Dingman is released. This occurs because the Dingman blade and teeth compress the endotracheal tube. A needle cap is placed alongside the endotracheal tube, and the Dingman is reopened. The tube will then be stented open. The lateral Dingman retractors are engaged to help the assistant see the operative field. A red rubber catheter is inserted through a nostril into the posterior pharynx. Then 2.0 silk is used to attach the posterior soft palate/uvular region to the tip of the catheter (Fig. 23-11, A). Later in the surgery, this will allow retraction of the soft palate to visualize the superior aspect of the surgical field. The bed is elevated to allow adequate visualization of the inferior posterior pharynx (distal extent of the donor sites). An assistant will provide upward traction on the Dingman handle to further visualize the inferior operative field.

Before an incision is made, the surgeon assesses through visualization and then manual palpation the presence of medially displaced internal carotid arteries. This is a rare phenomenon, seen in VCF, and if found, the surgery may continue, but with adaptation to avoid these structures. Bilateral palatopharyngeus muscle-containing myomucosal flaps will be elevated. Using a toothed forcep, the surgeon grasps the posterior tonsillar pillar, including mucosa and the muscle mentioned previously. With experience, the proper ratio of mucosa to muscle can be grasped (see Fig. 23-11). Using the pinpoint monopolar cautery on a blended cutting current, the myomucosal



flap harvest begins. The flap assumes the shape of a long pennant. The original grasp of the proposed flap is not released until enough release has been achieved to maintain a consistent ratio of muscle to mucosa. The optimal flap length will allow transposition without tension, with the tip reaching the contralateral lateral wall. To obtain this type of length, the flap is harvested inferiorly along the lateral pharyngeal wall about as far as a Bovie extender will allow. The flap should be elevated to the level of the adenoid pad. An assistant can use a Hurd tonsillar retractor on the lateral pharyngeal wall to facilitate accurate flap elevation. After each flap is elevated, the donor site is closed with 4-0 Vicryl. The soft palate is retracted using the attached red rubber catheter to allow upper field visualization. A transverse incision is made high in the posterior pharyngeal mucosa (typically at the adenoid pad), with some separation of the upper and lower edges, to allow both flaps to occupy the space. The superior pharyngeal constrictor muscle is not incised if at all possible to maintain the inset height of the flaps. The medial vertical myomucosal flap harvest incisions must connect to the horizontal pharyngeal mucosal incision, creating the upper part of a rectangle (Fig. 23-11, *B* through *E*). (This prevents lateral fistulas.) The lateral flap harvest incisions are tailored to release tension as the flaps are transposed.

The next portion of the operation focuses on flap transposition and inset. The first flap (either one) is transposed along the upper raw surface of the horizontal inset incision. Sequential 4-0 Vicryl sutures are placed from the flap base to the tip, attaching the superior mucosal edge to the edge of the adenoid pad. The suture bites through the adenoid pad should be deep, because this tissue is quite friable. The tip of the flap should insert at the junction of the posterior and lateral pharynx. Next, the lower flap is transposed. Again from the base to the tip, the flap is attached to the lower raw edge of the posterior pharyngeal incision. On completion, the tip of this flap should rest at the base of the previously inset flap. The final inset consists of sewing the mucosa of the two flaps together, centrally. The sphincter will be approximately 1 to 1.5 cm. The surgery is completed by releasing the red rubber catheter from the palate and suctioning the stomach with a nasogastric tube. Once the palate is released to its normal position, the flaps cannot be seen. The patient is awakened from anesthesia and extubated.

Discussion

Our preferred method for sphincter pharyngoplasty, as described previously, is a modification of the Hynes procedure, originally described in 1950, further refined by Jackson.³⁴ Orticochea³⁵ also described a version of this operation, but with lower inset of the flaps and limited lateral incisions. The modified Hynes approach allows inset of the sphincter at a more anatomic location with reference to the velopharyngeal mechanism and should prevent lateral fistulas. Some variance and debate still exists regarding flap elevation and inset. One point of variance is the amount of palatopharyngeus muscle harvested in each flap. Most authors espouse taking a good amount of muscle bulk, but how much is really necessary is difficult to quantify. Over the course of his career, Hynes recommended an increasing amount of muscle capture within the myomucosal flaps, including palatopharyngeus, salpingopharyngeus, and even some superior pharyngeal constrictor. A corollary to this is whether the muscle actually functions, contributing dynamically to velopharyngeal closure. The consensus seems to be that the muscles can be functional.^{36,37} Furthermore, what is the optimal size for the sphincter? End-to-end flap apposition has been described.³⁸ Complete overlap of the myomucosal flaps is generally advocated today.³⁴ Is there a way to predict accurately? Some authors recommend splitting the palate for further exposure in the superior aspect of the operative field.³⁸ Is this necessary given the retraction method described previously? Does this maneuver harm the long-term function of the soft palate?

Another area of variance in sphincter pharyngoplasty surgery is the location of the flap inset. Hynes's original description described inseting the flap at the site of velopharyngeal closure. Or-

ticochea's modification³⁵ directed flap inset at a much lower level. Based on a review study, high flap inset leads to better speech outcomes: VPD resolved in 93% of VPD patients who had a high flap inset versus 62% of lower flap inset patients.³⁹ Pigott⁴⁰ corroborated this finding, concluding that the flap should be inset as high as technically possible to bring it into the range of the normal level of the velopharyngeal mechanism.

One of the concerns of any speech operation that reduces the airway dimension is OSA. Nearly all patients undergoing this surgery will have a peak snoring phase of about 2 to 3 months postoperatively, with partial or complete resolution thereafter. Sommerlad²¹ showed mild to moderate OSA using polysomnography after sphincter pharyngoplasty (average patient age 9.3 years, range 3.6 to 18.9 years). Given this range, differences may exist in younger versus older children with regard to OSA after speech surgery.

Finally, is there a role for tonsil/adenoid management in patients who undergo sphincter pharyngoplasty? We advocate tonsillectomy 2 months preoperatively in patients with moderate to large tonsils, as have other investigators.²⁹ Conducting the operation and predicting speech results is much easier in this case. Can tonsils be reduced or removed *during* sphincter pharyngoplasty? Pang and Woodson⁴¹ reported simultaneous tonsillectomy and sphincteroplasty, but this procedure was for OSA.

Pharyngeal Flap

Positioning and Marking

Patient positioning is similar to that described previously for sphincter pharyngoplasty. The patient is prepared and draped in a sterile fashion. A marking pen may be used to outline the superiorly based flap on the posterior pharyngeal mucosa, the base of which rests at the level of the velum (Fig. 23-12, *A* through *F*).

Technique

A Dingman mouth gag may be opened, with tooth retractors in the middle (for full excursion). The device is inserted to the second level of tooth retractors, allowing better posterior pharyngeal visualization. (See the description of sphincter pharyngoplasty for information on managing a compressed endotracheal tube.) As in a sphincter pharyngoplasty procedure, before making surgical incisions, the surgeon visually and manually searches for medialized internal carotid artery or arteries (seen in VCF). With experience, surgeons can estimate proper flap width, but in general, wider is better. Based on a rough estimate, at least two thirds of the posterior pharynx is harvested. Visualization of the anticipated lateral port size further aids in creating the flap width. The surgeon and the assistant on the opposite side grasp the uvula (and/or the soft palate midline). Using a No. 11 blade, the soft palate is split, full thickness, in the midline anteriorly to the hard-soft junction. The flap harvest is initiated. The assistant lifts the handle of the Dingman upward to allow a view deeper into the inferior posterior pharynx. Using blended cutting currents with monopolar cautery, flap elevation is initiated, including mucosa and superior pharyngeal constrictor muscle. Visualization of the prevertebral fascia during the dissection indicates appropriate flap thickness. The flap is elevated superiorly until it can be transposed and contacts the caudal edge of the hard palate without tension. The lower portion of the donor site is closed with 4-0 Vicryl.

Bilateral nasal mucosal pennant flaps are elevated. The posteromedial cut edge of the soft palate is grasped with a single hook and reflected laterally, exposing the nasal mucosa. The nasal mucosa is incised to create a triangle with the base (widest part) along the posterior soft palate. The delineated triangle flap is elevated so that it transposes posteriorly, containing mucosa and subcutaneous fat. This is done bilaterally. These flaps will cover the raw surface of the pharyngeal flap after the initial inset.

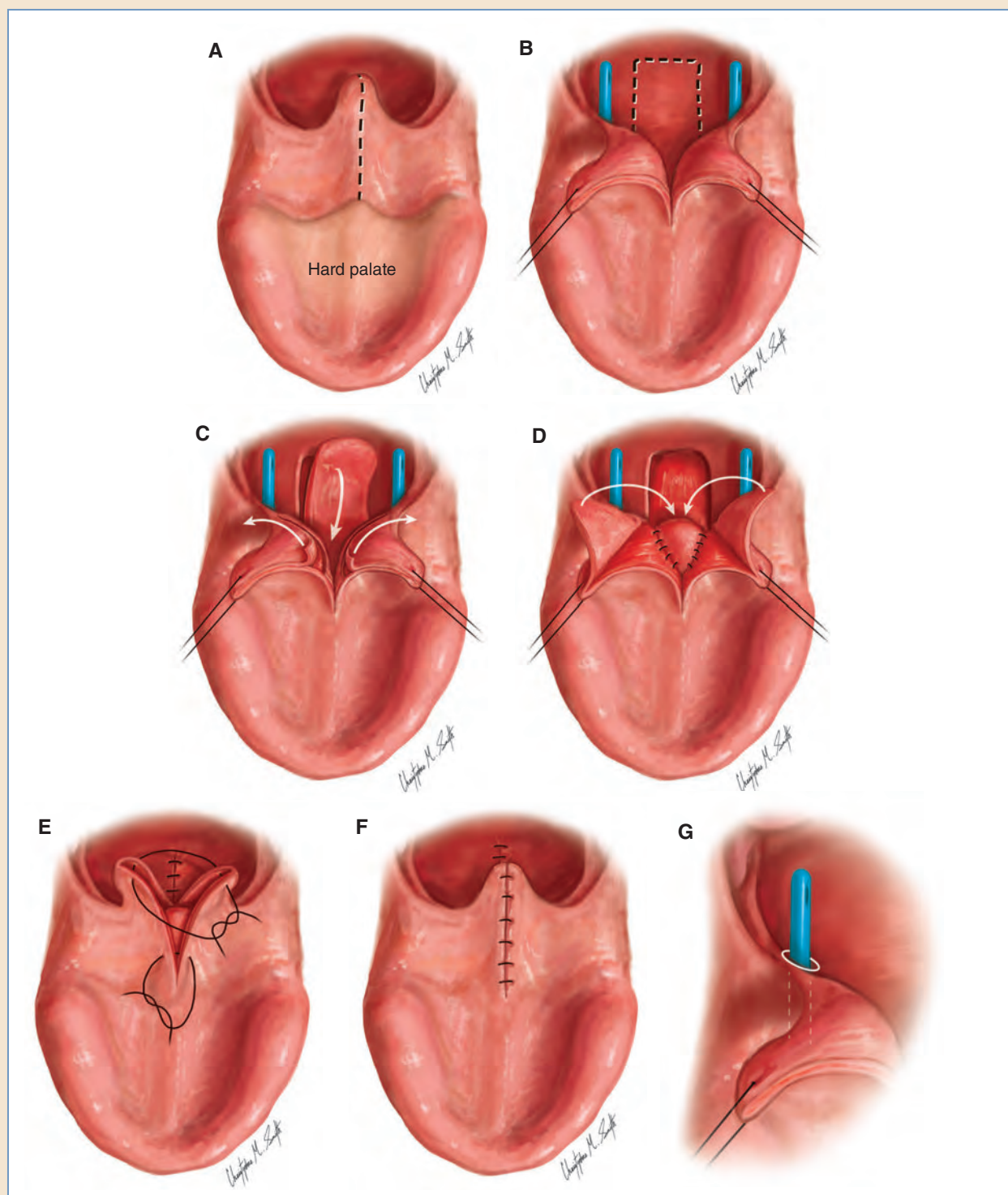


Fig. 23-12 Steps of superiorly based posterior pharyngeal flap. **A**, Soft palate is opened in midline from uvula to posterior hard palate. **B**, Catheters will be used to gauge lateral “port” size. Soft palate is retracted. The superiorly based posterior pharyngeal flap is marked, to include mucosa and superior pharyngeal constrictor muscle. **C**, Bilateral nasal mucosa pennant flaps are elevated and the posterior pharyngeal flap is brought into the soft palate. **D**, The pharyngeal flap has been inset. The nasal mucosa pennant flaps are transposed to cover the raw surface of the transposed pharyngeal flap. **E**, Final oral and nasal mucosal closure. **F**, The donor site is closed primarily. **G**, The use of 14-gauge red rubber catheter (shown in blue) to modulate port size.

The flap is inset and the port size contoured. Using mattress sutures of 4-0 Vicryl, the flap inset begins in the midline near the hard-soft palate junction. This is the last opportunity to adjust flap tension. Progressing laterally, from the tip of the flap toward its base, sequential mattress sutures are placed along each side of the flap to the lateral soft palate to secure the flap inset. Bilateral 14 Fr red rubber catheters are placed transnasally and in each lateral pharyngeal location (Fig. 23-12, G). The catheters do not indicate the final port size, but aid in visualization and overall port size adjustment. As the sequential flap mucosa to lateral soft palate nasal mucosal sutures are placed, a general idea of port size can be determined. Most of the port size is created at this point, where visualization is better. The final port size can be adjusted after the lining is closed. This varies from patient to patient; however, 1 to 1.5 cm is a rough estimate. The previously raised bilateral soft palate nasal mucosal pennant flaps are transposed. The key suture is placed to unite the tips of the flaps and to anchor them to the base of the flap (at the posterior pharyngeal wall). The flaps are closed distal to proximal, grasping a bite of flap muscle to create an adherence. Closure of the midline to the hard-soft junction is completed. The final lateral port adjustments can be made. The flap donor site is closed, up to the base of the transposed flap. If postoperative airway obstruction is a concern, the surgeon can place a nasopharyngeal airway (or two). The stomach is suctioned with a nasogastric tube, and the Dingman is removed. The patient is then awakened from anesthesia and extubated.

Discussion

The pharyngeal flap surgery detailed previously is a modification of the superiorly based pharyngeal flap, originally described by Schönborn in 1876⁴² and popularized by Padgett.⁴³ The goal of this procedure is a midline subtotal airway obstruction, with improved velopharyngeal function, without airway compromise. Several aspects of this procedure have been and continue to be discussed.

First, should the flap be inferiorly or superiorly based? The literature on this issue is mixed. Proponents of an inferiorly based flap claim that scarring in the posterior pharynx will have a cephalad pull (because of the orientation of the raw surface on the nasal aspect of the flap, pulling it up into a more optimal position with the velopharyngeal mechanism).⁴⁴ A disadvantage is length limitation. A superiorly based flap can be long and tailored to the port size as needed. Many studies have shown function is equivalent; however, Wattanawong et al⁴⁴ showed improved correction of VPD with an inferiorly based flap with a head-to-head comparison.

Another area that continues to prompt discussion is the proper flap width. Many advocate making the widest flap possible (without creating complete airway obstruction). Shprintzen et al⁴⁵ recommended tailoring the width of the flap based on the amount of lateral wall movement during a nasopharyngeal examination. Others have supported this idea.⁴⁶ Although this concept is appealing theoretically, accurate practical application in the operating room can be difficult. Another approach to flap width is a "short wide flap." Some think this type of flap is less prone to inferior scarring and can truly create a subtotal obstruction of the nasopharynx, an outcome potentially desirable in patients with an adynamic velopharyngeal mechanism such as those with VCF.

A further refinement of superiorly based pharyngeal flap surgery is the lateral port control. Hogan⁴⁷ is credited with the method for adjusting port size. Based on findings from prior studies on oropharyngeal airway flow dynamics, he recommended controlling lateral port size during surgery with a 4 mm² catheter placed in each nasal orifice. These studies showed an increase in nasal airflow between 10 to 20 mm² and frank nasal air escape of more than 20 mm². The specific aspect of superiorly based pharyngeal flap surgery that controls the lateral port size is the use of soft palate nasal mucosal pennant flaps. When sequentially sutured to the lateral aspect of the pharyngeal flap, the mucosal pennant flaps can be used to progressively narrow the lateral port orifice.

The early descriptions of posterior pharyngeal flap surgery did not include reference to coverage of exposed raw surfaces during surgery.⁴⁸ Since that time, refinements to this surgery have included closure of the donor site, which decreased postoperative pain and scarring. With the use of an unlined pharyngeal flap, practitioners noted undue horizontal contracture, which predisposed to postoperative continuance of VPD. Bilateral nasal soft palate pennant flaps provide mucosal lining to the raw surface on a superiorly based flap, thus decreasing uncontrolled contracture.⁴⁹

For many years the pharyngeal flap has been considered the benchmark for surgical correction of VPD. However, it has also been known to place the patient's airway at risk, both in the immediate postoperative period and in the long term, with the potential for OSA. Most patients will snore in the immediate postoperative period, because of both the flap itself and early postoperative swelling. Usually by about 3 months after surgery, most or all of the snoring will resolve. However, the long-term airway findings are most important. Liao et al⁵⁰ used polysomnography to assess the risk of OSA in adults and children after pharyngeal flap surgery. They found a 93% prevalence of some level of OSA more than 6 months after surgery, with 58% of patients in the moderate to severe category. Flap width was not an independent variable leading to the development of OSA. This rate of OSA is much higher than in many of the prior studies.⁴⁹⁻⁵¹ Lesavoy et al⁵² showed a 38% rate of airway obstruction based on an observational obstruction scale and a limited sleep study analysis in the early postoperative period. Over time the obstructive symptoms subsided and 2 of 32 patients had severe OSA on polysomnography after 3 months. The authors concluded it was necessary to accept transient airway obstruction to achieve velopharyngeal competence. Ysunza et al⁵³ reported on a series of 571 patients who underwent superiorly based pharyngeal flap procedures. Eighteen had obstructive symptoms, with 15 (2.6%) scoring *severe* on polysomnography.

There is currently no consensus on the severity of long-term airway obstruction in pharyngeal flap surgery. One difficulty is the variability with which this outcome has been studied. A polysomnogram performed several months after surgery and scored by a trained pediatric sleep specialist would be the benchmark for measuring long-term airway impairment. Very few studies have achieved this level of detail.

Another area of debate regarding pharyngeal flaps is whether the flap is dynamic in velopharyngeal functioning. Most agree that the most significant aspect of functioning within the velopharyngeal mechanism in patients who have had a pharyngeal flap is the lateral wall motion. However, Broadbent and Swinyard⁵⁴ performed an electromyographic study showing variable amounts of muscle activity within posterior pharyngeal flaps.

Pharyngeal Augmentation

Alloplastic Materials

A subset of patients with mild VPD and a small gap on velopharyngeal closure have unsuccessful speech therapy. The size of the gap does not substantiate the use of Furlow lengthening, sphincter pharyngoplasty, or a pharyngeal flap. A variety of methods of posterior pharyngeal wall augmentation have been employed to treat this particular group of patients. Several alloplastic materials have been used, including silicone, Teflon, polyethylene, and Proplast. Data are mixed regarding the efficacy of these materials in treating VPD.⁵⁵ Issues have been reported of caudal migration of implants along the prevertebral fascia, which nullifies the functionality of the implant. As with any prosthetic material, extrusion and infection are risks. A study on the use of Proplast I showed promising results for correction of VPD, with a reasonable safety profile.⁵⁶ Another promising method using alloplastic material is the injection of calcium hydroxylapatite (Radiessce). A review study showed good tolerance of the material with improved velopharyngeal function.⁵⁷

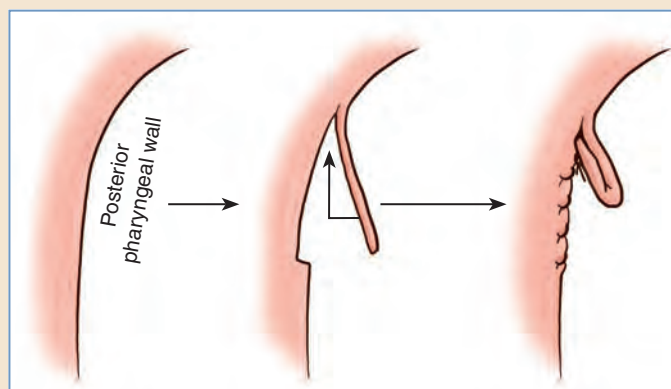


Fig. 23-13 Posterior pharyngeal augmentation with a rolled flap.

Autologous Materials

Several methods involving autologous material have been used for VPD. Autologous fat injection has showed mild improvement in VPD; however, it is prone to resorption. Rolled dermis inserted in to the Passavant ridge region has had a similar profile. Last, posterior pharyngeal augmentation with a rolled flap has been evaluated. A superiorly based posterior pharyngeal flap is created and rolled on itself (Fig. 23-13). The base is sutured near the tubercle of the atlas. This flap has not been shown to compromise the airway, but it has not led to notable speech outcome improvement.⁵⁸

POSTOPERATIVE CARE

Patients who have had speech surgery must be treated as having a delicate airway. Swelling, bleeding, and oral secretions may be difficult for the patients postoperatively. They require at least overnight monitoring in a step-down unit or a unit that can provide continuous oxygen monitoring. Some patients need a nasal airway placed carefully and under direct vision by the surgeon in the operating room for temporary maintenance of the airway until they are more awake. Occasionally, it is useful to suture this nasal airway to the septum to prevent it from accidental dislodgement. IV fluids are continued until patients are able to drink adequately. Hard foods and other hard objects should not be placed in the mouth. The patient is limited to a strict liquid and pureed diet for the next 3 to 4 weeks postoperatively. Pain is controlled with acetaminophen and ibuprofen alone, and narcotic pain medications are given for breakthrough pain. Having all medications in a liquid oral formulation is helpful. Patients can be discharged home on the first postoperative day if their pain is controlled, they are drinking, urine output is adequate, and they have had no untoward events noted on their continuous oxygen monitors. A routine follow-up is scheduled for 2 weeks postoperatively. Parents and patients are counseled that speech may worsen before it improves. Patients may initially snore before the swelling improves.

Postoperative Speech Therapy

Postoperative speech therapy is delayed until the velopharynx recovers from surgery, and the surgeon deems it to be healed enough to be stressed by speech therapy. Typically, this occurs

approximately 6 weeks after the procedure. Parents are counseled that their child's speech may not be improved immediately, but some tonal difference should be noted initially. As swelling continues to improve, and as patients are able to participate in more active speech therapy, it is generally expected that speech will improve over time.

TREATMENT OUTCOMES

Several retrospective studies and some prospective analyses on the outcomes of sphincter pharyngoplasty and pharyngeal flap have been conducted. Sie et al⁵⁸ retrospectively studied patients with repaired cleft palates and syndromic diagnoses who underwent sphincter pharyngoplasty for VPD. Twenty of 24 (83%) had complete or significant resolution of VPD on perceptual speech assessment, and six (25%) had residual VPD. Of these six patients, three had a secondary surgery and complete resolution of VPD. In cases of failed sphincter pharyngoplasty, the cause appears to be related to the level of insertion of the myomucosal flaps. Flaps inserted too low on the posterior pharynx (lower than the CI–adenoid pad region) tend to result in inadequate velopharyngeal closure postoperatively.⁵⁹ Others have studied the ability of the sphincter pharyngoplasty to achieve dynamic function, concluding that nearly all newly created sphincters will have dynamic function, but movement of the sphincter has no influence on postoperative speech outcomes.^{37,38}

Pharyngeal flap surgery has been effective in treating VPD in an isolated retrospective analysis. In a study of 60 patients with a pharyngeal flap,⁵⁴ 78% achieved normal speech, 18% were hyponasal, and 3% had persistent hypernasality.⁴⁹ Pharyngeal flap surgery has been shown to be effective in syndromic patients as well. In a retrospective analysis of patients with VCF with VPD who had a pharyngeal flap, Swanson et al⁶⁰ found an 88% correction rate, with 1 of 33 showing signs of sleep apnea.

Multiple studies have compared sphincter pharyngoplasty and pharyngeal flap surgery for speech outcomes. The VPD Surgical Group reported on a large, multiinstitutional randomized trial of sphincter pharyngoplasty and pharyngeal flap for treating secondary VPD.⁶¹ They found no difference in speech outcomes. This work corroborated earlier studies, including a large series (168 patients) that showed similar outcomes for speech correction (70% to 75%) with either technique.^{62,63} However, the comparison studies showed a trend toward increased airway obstruction in pharyngeal flap patients, compared with sphincter pharyngoplasty patients, and a small percentage chance of overcorrection (hyponasality).

Complications

The basic goals of speech surgery are to improve, if not normalize, VP function and prevent long-term OSA. Noted complications of Furlow palatoplasty, sphincter pharyngoplasty, and pharyngeal flap revolve around these two goals. In the acute setting, bleeding can occur. Most postoperative bleeding is self-limited but can require returning the patient to the operating room if it is persistent and/or threatens airway function. The health care team should be vigilant to ensure that patients do not develop an acute airway obstruction in the immediate postoperative period and are monitored in an intermediate care setting. Pharyngeal flap patients (and Furlow palatoplasty patients) are most at risk for this impairment. An intraoperatively placed nasopharyngeal airway can ameliorate this problem. Subacute complications include dehiscence and fistula formation. Long-term speech complications include persistent velopharyngeal dysfunction, as evidenced by hypernasality or hyponasality. After about 3 months, patients should be evaluated for symptoms and signs of persistent airway obstruction. They may require polysomnography to rule out OSA if indicators are present, such as persistent snoring, daytime somnolence, irritability, and an inability to concentrate.

FUTURE DIAGNOSTIC AND TREATMENT MODALITIES

Robotic Surgery

The Children's Hospital of Philadelphia recently conducted a feasibility study wherein they used cadaver heads and a robotic telemanipulator to perform pharyngeal flaps.⁶⁴ They noted the learning curve was steep, but the setup was ergonomic. Visibility of the ports was simplified by the use of a 30-degree endoscope. In the future, with improving robotic technology it may be possible to perform these technically demanding operations under better ergonomic circumstances with better visualization.⁶⁴

Magnetic Resonance Imaging

Dynamic MRI of the velopharynx may be reformatted to simulate endoscopy. This technology has not been used in a widespread fashion yet, but may be helpful in the future as a type of virtual endoscopy or as an adjunct procedure.

KEY POINTS

- A multidisciplinary approach is best for patients with VPD.
- VPD may have either structural or neurologic causes.
- Orofacial cleft patients with a history of a repaired cleft palate are the most common group to present with VPD in school-age children.
- Patients with a VCF abnormality may have aberrant carotid artery anatomy.
- A palpable notch at the junction between the hard and soft palate, a zona pellucida, and a bifid uvula may be signs of a submucous cleft palate.
- Both sphincter pharyngoplasty and a pharyngeal flap carry a risk of OSA, with a greater risk with pharyngeal flap.
- A comprehensive examination and speech workup should be completed by an interdisciplinary team that includes a surgeon and an SLP before speech surgery is considered.
- Patient diagnosis, the severity of VPD, and findings on NP examination drive the selection of the speech surgery procedure.
- Superiorly based pharyngeal flap use is most popular, but the literature supports using either an inferior- or a superior-based flap for treating VPD.

Continued

KEY POINTS (continued)

- The pharyngeal flap width can be tailored based on the amount of lateral wall motion on nasopharyngeal examination.
- With good palate excursion and poor lateral wall motion, sphincter pharyngoplasty is a good treatment for VPD.
- In patients who have had a sphincter pharyngoplasty, myomucosal flaps have been shown to be dynamic.
- Surgical inset of transposed palatopharyngeus flaps in sphincter pharyngoplasty should be at the level of the C1-adenoid pad.
- Most secondary VPD is nonsyndromic and related to a history of cleft palate repair, and the next most common cause is a 22q deletion.

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24

Craniosynostosis

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To the inexperienced, craniosynostosis might seem like a kaleidoscope of unusual craniofacial dysmorphology managed only by craniofacial surgical specialists. However, it can be defined more simply as a premature fusion of one or more cranial sutures that leads to characteristic and specific deformities of the cranial vault. It can occur in the postnatal period, but most cases develop in utero and may be diagnosed at birth. Craniosynostosis can be an isolated event or associated with a syndrome.

Functional implications of craniosynostosis are related to the dynamic growth of the craniofacial skeleton and depend on the number of involved sutures and the presence of a syndrome. However, in isolated cases, they focus primarily on neurologic and psychosocial issues related to the aesthetic deformity. Goals of surgical treatment are to improve shape, form, and function and to expand cranial vault volume to accommodate ongoing brain growth.

Management is multidisciplinary, provided by a skilled team of specialists. It should be the domain of tertiary or quaternary care centers with high volumes. This chapter discusses implications and management options for patients who present with craniosynostosis.

HISTORICAL AND ETIOLOGIC ASPECTS

Although several Greek and Roman authors have referred to the condition of craniosynostosis, the pathology was not recognized until Vesalius¹ published *De Humani Corporis Fabrica Libri Septem* in 1543 (Fig. 24-1). This publication provided clear illustrations of brachycephalic and plagiocephalic skulls.

In 1830 Otto² proposed that a consequence of premature suture fusion was compensatory cranial expansion along another trajectory and was the first to use the name *craniosynostosis*. However, the bulk of the credit went to Virchow,³ a German anatomist at the turn of the century, who described *Virchow's law*. This essentially states that the observed deformities occur as a result of “cessation of growth across a prematurely fused suture,” with “compensatory growth” along non-fused sutures in a direction parallel to the affected suture, causing obstruction of normal brain growth (Fig. 24-2). This law is a very useful tool for understanding and establishing the diagnosis of craniosynostosis.

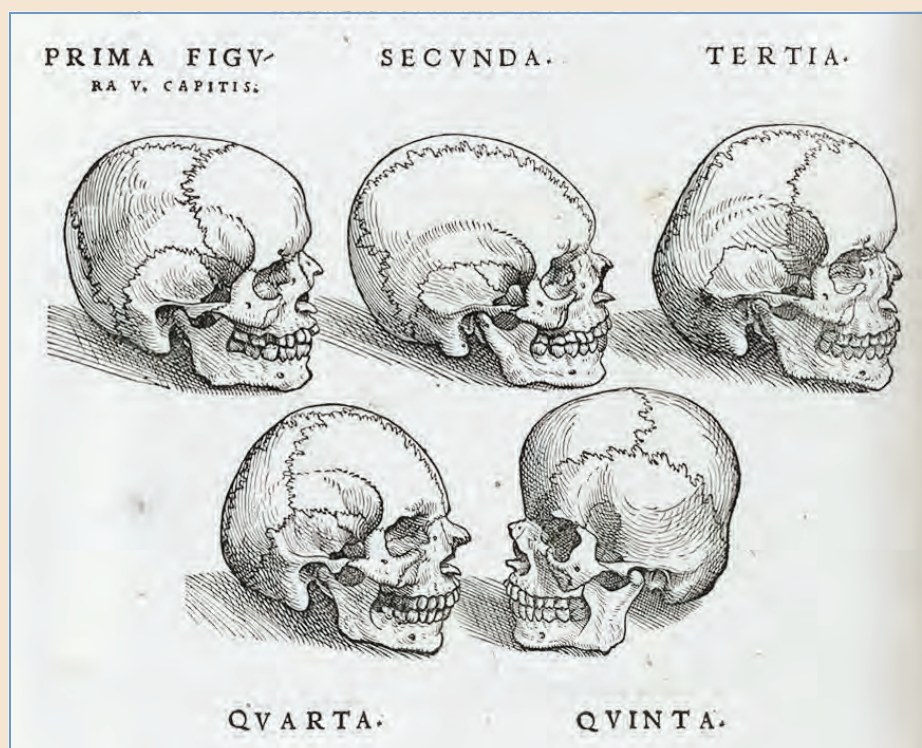


Fig. 24-1 Image from Vesalius' *De Humani Corporis Fabrica Libri Septem* (published in 1543) demonstrating cranial dysmorphology caused by craniosynostosis. (Courtesy of the British Library.)

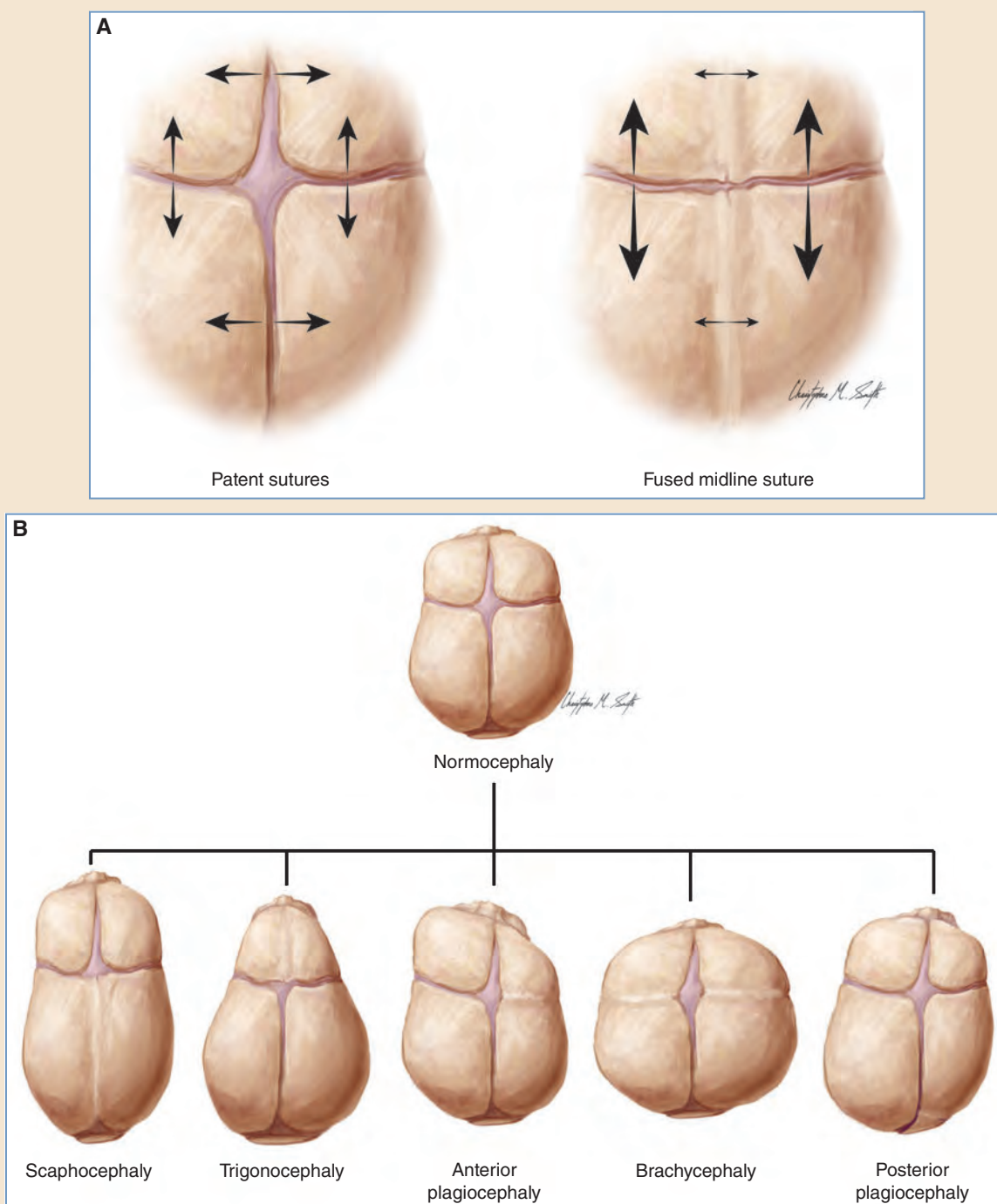


Fig. 24-2 Virchow's law of skull growth. **A**, The cranial sutures allow expansion and growth of the skull at right angles to the sutures (*left*). When a suture is fused, bone expansion ceases in a direction perpendicular to the fused suture, and compensatory expansion occurs in the opposite direction because of the presence of the patent sutures (*right*). **B**, This leads to predictable deformities of the calvarial vault that are diagnosed as single-suture craniosynostosis in a clinical examination. Cases of multiple-suture involvement and syndromic craniosynostosis are more challenging and require radiographic investigation.

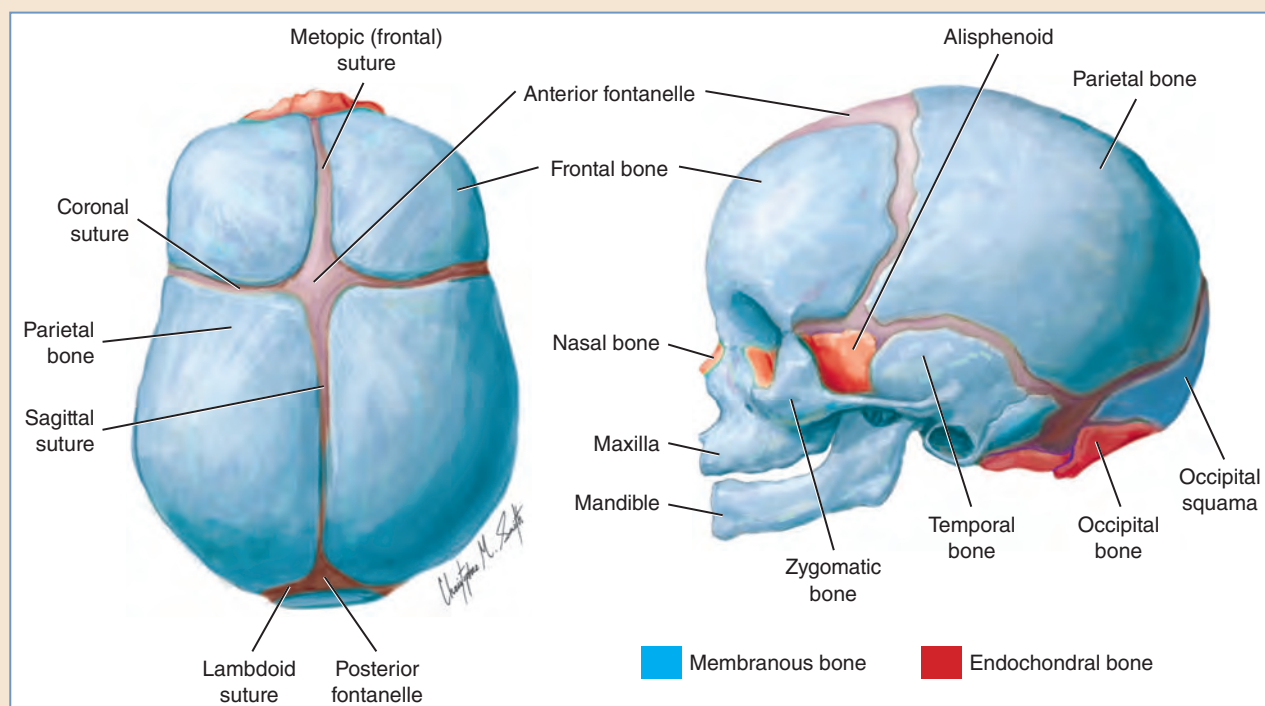


Fig. 24-3 Embryology of the craniofacial skeleton. Endochondral bone makes up most of the skull base, and membranous bone forms the cranial vault.

Embryology

The skull is a unique and dynamic structure composed of the neurocranium (brain case) and viscerocranium (bones of the lower face) that appear embryologically at 6 to 8 weeks and begin to ossify by 12 weeks' gestation (Fig. 24-3). The components of the neurocranium are classified as *cartilaginous* (skull base) and *membranous* (skull plates), according to the method of bone formation. Endochondral ossification occurs after the development of a cartilaginous precursor and forms the sphenoid, occipital, and petromastoid portion of the temporal bone and ethmoid bones. Intramembranous ossification occurs from a layer of mesenchyme under the dermal mesenchyme and above the meninges as a series of centers that coalesce radially to form the parietal, frontal, and squamous portions of the temporal bones, with input into the frontal bone by neural crest cells. Sutures form between the ossifying plates of bone, and fontanelles (soft spots) form where the three sutures come together.

Anatomy

In infants, the calvarial vault comprises two parietal bones, two frontal bones, two temporal bones, and the occipital bone. These are joined by four major sutures (coronal, metopic, sagittal, and lambdoid) and multiple minor sutures (squamosal, sphenofrontal, sphenosquamosal, sphenoparietal, parietomastoid, and occipitomastoid) (Fig. 24-4). The occipital and temporal bones are compound bones with multiple ossification centers that indicate fusion of more than one embryonic element. Occasionally, extra bones form within sutures known as *wormian* or *intrasutural*

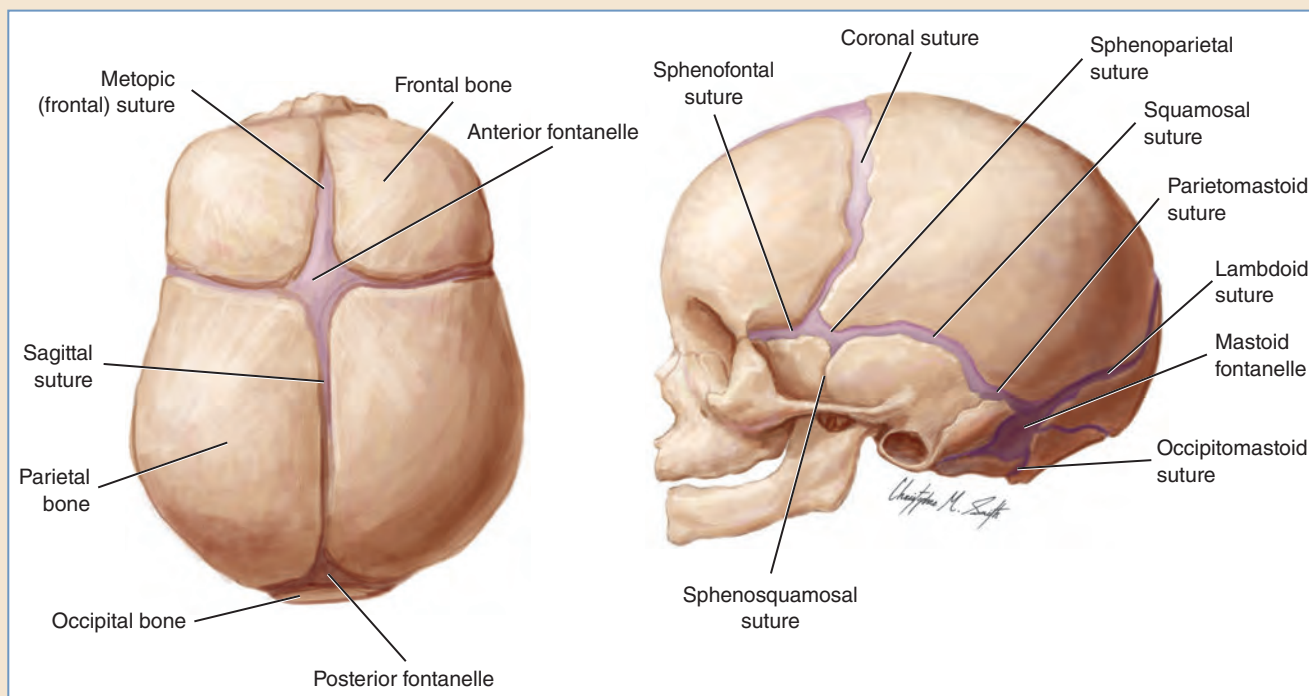


Fig. 24-4 The anatomy of an infant skull, showing the major sutures, the minor sutures, and the fontanelles.



Fig. 24-5 This CT scan shows the presence of wormian (intrasutural) bones in the lambdoid suture.

bones (Fig. 24-5). These irregular, isolated bones often arise in the course of the lambdoid suture and may be a marker for numerous diseases. Major landmarks of the skull are the bregma, which is the point of intersection between the sagittal and coronal sutures, and the lambda, which is the posterior point of intersection of the sagittal and lambdoid sutures.

Cranial sutures are a type of fibrous joint (synarthrosis) that allows small amounts of movement and contribute to the elasticity and compliance of the skull. This facilitates molding of the head during passage through the birth canal and rapid growth of the skull in the first few years of life. Sutures are signaling centers and regulate the balance between proliferation and differ-

entiation of osteogenic precursor cells during the first few years of life. Cartilaginous synchondroses are the equivalents on the skull base but are formed differently because of endochondral ossification.

The human calvarial vault has six fontanelles, which form where three sutures come together (see Fig. 24-4). They consist of two unpaired (anterior and posterior) and two paired (anterolateral or sphenoid and posterolateral or mastoid) fontanelles. The fontanelles are nonossified membranous areas that allow rapid stretching and deformation of the cranium as the brain expands. The posterior fontanelle usually fuses by 3 months of age, and the anterior fontanelle is the last to close, usually by 18 months of age. The fontanelles can bulge with underlying intracranial pathology such as increased pressure. Large, confluent fontanelles may be seen in craniofacial dysostosis syndromes. However, absent or small fontanelles are not necessarily a cause for alarm in the absence of other pathology or developmental delay.

Growth and Development

According to the functional matrix hypothesis proposed by Moss and Salentijn,⁴ bones do not grow but are grown: “form follows function.” This dictates that the growth of the skeleton is a secondary response to the demands of the related functional matrices, which, in the case of the calvarium, is very much influenced by the growth of the brain. Human brain growth is robust, tripling in size by 1 year of age, and reaches 85% of adult size by 3 years of age.⁵ Primary growth of the brain results in a stimulus for secondary growth of the sutures and synchondroses, with resultant deposition of bone on the sutural edges. This is the predominant form of calvarial growth until about 4 years of age, when the head circumference is approximately 90% that of an adult size. Subsequent growth of the skull is less dependent on sutural expansion and is appositional in nature, with bone deposited on the ectocranial and endocranial surfaces, resulting in an increase in bone thickness and expansion of the medullary space (and formation of the diploe). Cranial sutures fuse in a predictable manner from front to back and from lateral to medial, with the metopic suture closing by 8 to 9 months of age.⁶ The remaining sutures are patent until the third decade of life, and they fuse in the following sequence: sagittal suture (22 years of age); coronal suture (24 years of age); lambdoid suture (26 years of age) and squamosal suture (after 60 years of age).

CLASSIFICATION FOR CRANIOSYNOSTOSIS

Myriad Greek terms are used to describe the head shapes associated with craniosynostosis but do not provide a useful methodology for classification. Craniosynostosis is best categorized according to the number of involved sutures (single or multiple), the anatomic location (sagittal, metopic, unilateral or bilateral coronal, lambdoid, or minor sutures), and whether it is associated with a recognized syndrome (Box 24-1). Craniosynostosis has been recognized as a component of more than 180 syndromes, but the most common are Apert, Crouzon, Pfeiffer, Saethre-Chotzen, and Muenke syndromes.

EPIDEMIOLOGY, NOMENCLATURE, AND INCIDENCE

Craniosynostosis has an incidence of 1 in 2000 to 1 in 5000 newborns.^{7,8} Most (85%) are categorized as single-suture and nonsyndromic. The suture most often involved is the sagittal suture, causing sagittal synostosis. More recently, it appears that metopic synostosis has displaced unilateral coronal synostosis (range 1 in 10,000 to 1 in 15,000 live births) as the second most common form in Europe and the United States.^{9,10} Reported cases of lambdoid synostosis may have been overestimated, because posterior plagiocephaly may actually have been attributed to deforma-

Box 24-1 Classification of Craniosynostosis**Anatomic****Single-suture**

Sagittal
Metopic
Unicoronal
Lambdoid
Minor suture (frontosphenoid,
zygomaticotemporal)

Multiple-suture

Any combination possible, usually bicoronal

Cause**Primary**

Nonsyndromic
Scaphocephaly (sagittal)
Trigonocephaly (metopic)
Anterior plagiocephaly (unicoronal)

Posterior plagiocephaly (lambdoid)
Turribrachycephaly (bicoronal)
Oxycephaly (delayed bicoronal)
Syndromic (more than 180 syndromes with
craniosynostosis)
Apert syndrome
Crouzon syndrome
Pfeiffer syndrome
Jackson-Weiss syndrome
Carpenter syndrome
Muenke syndrome
Saethre-Chotzen syndrome

Secondary

Biomechanical (shunt)
Bone metabolic disorders
Nutritional
Oxycephaly

Table 24-1 Classification and Incidence of Craniosynostosis

Name	Shape	Etiologic Factors	Incidence
Scaphocephaly	Keel- or boat-shaped	Sagittal synostosis	1 in 2500 ^{7,8}
Dolichocephaly	Long and narrow		
Trigonocephaly	Triangular	Metopic synostosis	1 in 10,000 ^{9,10}
Plagiocephaly	Asymmetrical, anterior or posterior flattening	Unilateral coronal synostosis Lambdoid synostosis	1 in 15,000 ^{9,10} 1 in 100,000 ^{11,70}
Turriccephaly	Tall or tower	Bilateral coronal synostosis	Variable with syndromes
Brachycephaly	Short, broad		
Turribrachycephaly	Tall, short, broad		
Oxycephaly	Conical or pointed skull, delayed-onset	Bilateral coronal synostosis	Rare
Kleeblattschädel	Cloverleaf, trilobar	Pansynostosis	Rare

tional forces. True lambdoid synostosis is rare, with an incidence of 1 in 100,000 live births.¹¹ In single-suture, nonsyndromic craniosynostosis, the symmetrical (sagittal and metopic) forms have a male predominance, and the asymmetrical (unicoronal) forms have a female predominance.

The nomenclature for the cranial deformities associated with craniosynostosis can be confusing (Table 24-1). The names are descriptive only; skull dysmorphism also can result from non-synostotic pathology (for example, scaphocephaly from prematurity).⁹

Syndromic craniosynostosis is relatively rare, with 1 in 25,000 to 1 in 100,000 infants affected.⁷ More than 180 different syndromes are associated with craniosynostosis^{9,12,13} (Table 24-2).

Table 24-2 Nomenclature of Craniosynostosis

	Apert Syndrome	Crouzon Syndrome	Pfeiffer Syndrome	Muenke Syndrome	Saethre-Chotzen Syndrome	Craniofrontonasal Syndrome
Gene mutation	<i>FGFR2</i>	<i>FGFR2</i>	<i>FGFR1</i> <i>FGFR2</i>	<i>FGFR3</i> : Pro250Arg ²⁰	<i>TWIST</i>	<i>EFNB1</i>
Inheritance	Autosomal dominant	Autosomal dominant	Autosomal dominant	Autosomal dominant	Autosomal dominant	X-linked
Incidence	1 in 70,000 ⁷⁸	1 in 65,000 ^{71,80}	1 in 100,000 ⁸²	1 in 30,000 ^{85,86}	1 in 25,000 to 1 in 50,000 ⁸³	1 in 100,000 to 1 in 120,000 ⁸⁸
Skull morphology	Bilateral coronal synostosis	Bilateral coronal synostosis	Bilateral coronal synostosis, cloverleaf skull in type 2 Pfeiffer	Unilateral or bilateral coronal synostosis, macrocephaly	Unilateral or bilateral coronal synostosis, metopic synostosis	Unilateral or bilateral coronal synostosis
Facial features	Proptosis, cleft palate, high-arched palate, partial adontia, midface hypoplasia	Proptosis, hypertelorism, strabismus, mandibular prognathism, midface hypoplasia	Proptosis, hypertelorism, supernumerary teeth, midface hypoplasia	Proptosis, hypertelorism, high-arched palate, mild midface hypoplasia	Lid ptosis, ear anomalies, low frontal hairline, midface hypoplasia	Hypertelorism, broad nasal bridge, bifid nasal tip; rarely, cleft lip and/or palate (females more severely affected), dry frizzy hair
Limb anomalies	Syndactyly (second through fourth digits), radiohumeral fusion, progressive tarsal bone fusion	Tarsal bone fusion, symphalangism	Broad toes and thumbs, tarsal bone fusion, elbow ankylosis, elbow synostosis, cutaneous syndactyly	Brachydactyly of hands and feet, symphalangism, Klippel-Feil cervical anomaly	Cutaneous syndactyly, hallux valgus, duplicated distal phalanx of the hallux	Asymmetrical lower limb shortness, broad hallux, joint laxity, cutaneous syndactyly, grooved nails
Neurocognitive impairment	Severe intellectual disability and/or developmental delay, conductive hearing loss	Intellect normal, conductive hearing loss	Intellect normal in Pfeiffer type 1, intellectual disability and developmental delay in Pfeiffer type 2 and 3, conductive hearing loss	Intellectual disability and/or developmental delay, low-frequency sensorineural hearing loss	Intellect normal; developmental delays common in those with gene deletions; conductive, mixed, and profound sensorineural hearing loss	Normal intellect in >50%, 10% to 50% with developmental delay, occasionally learning difficulties (mild), sensorineural hearing loss

Genetics

A clinical and molecular genetic assessment is a crucial part of managing infants who present with craniosynostosis. A genetic association was first reported in 1993 in a family cohort in Boston and was linked to a mutated copy of *MSX2*, a homeobox gene thought to function in inductive tissue interactions during embryogenesis and caused by a single amino acid substitution.¹⁴ Boston-type craniosynostosis is a highly penetrant autosomal dominant form of syndromic craniosynostosis that is rarely seen outside the original cohort, but its importance in demonstrating

the role of genetics cannot be overstated. Subsequently, mutations in the *fibroblast growth factor receptor* (*FGFR*) genes, *TWIST1* genes, and *ephrin B1* (*EFNB1*) genes were found to be responsible for most syndromic craniosynostosis cases, although the field is dynamic and new mutations are identified each year.^{7,12} The FGF family is composed of 22 highly conserved proteins that are important in regulating cell proliferation, differentiation, and migration. *TWIST1* is a transcription factor that plays an important role in specifying and maintaining cell identity. *EFNB1* is a protein-coding gene involved in cell adhesion and functions in the development of the nervous system. Chromosomal alterations are responsible for 10% to 30% of syndromic craniosynostoses.¹³ In contrast, nonsyndromic, single-suture synostoses appear to have less correlation with genetic mutation, though recent evidence suggests increased *RUNX2* expression may represent a transcriptomic signature for a subset of single-suture craniosynostosis cases.¹⁵

More than 180 craniosynostosis syndromes have been identified, with more than 60 different genetic mutations.¹⁶ Most of these mutations occur in *FGFR2*. Eight fibroblast growth factor receptor-mediated craniosynostoses have been reported:

1. Apert syndrome (*FGFR2*)
2. Crouzon syndrome (*FGFR2*)
3. Jackson-Weiss syndrome (*FGFR2*)
4. Muenke syndrome (*FGFR3*)
5. Crouzon syndrome with acanthosis nigricans (crouzonodermoskeletal syndrome) (*FGFR3*)
6. Pfeiffer syndrome (*FGFR1* and *FGFR2*)
7. Beare-Stevenson syndrome (*FGFR2*)
8. *FGFR2*-related isolated coronal synostosis

More than 100 distinct mutations in *TWIST1* have been identified as causative of Saethre-Chotzen syndrome.¹⁷ Craniofrontonasal dysplasia is caused by mutations in the *EFNB1* gene.

PATHOPHYSIOLOGY

Early theories of the pathophysiology of craniosynostosis included infection, metabolic conditions, biomechanical constraint, tensile forces between the cranial base and neurocranium, abnormalities in the cranial base, and primary pathology within the developing sutures. These have largely been supplanted by advances in genetics and sophisticated molecular research in animal models of both normal cranial suture fusion and knockout animals with craniosynostosis, resulting in improved understanding of molecular interactions in the suture complex.

Opperman et al¹⁸ and Roth et al¹⁹ have conducted landmark studies in tissue interaction. They demonstrated the integral role of dura mater in preventing synostosis, suggesting biochemical interactions rather than biomechanical forces were responsible for maintaining the suture. Molecular studies have highlighted the interconnection of transcription factors, cytokines, growth factor receptors, and extracellular matrix molecules. Most craniosynostosis syndromes result from mutations in genes encoding *FGFR1*, *FGFR2*, and *FGFR3*, in addition to the transcription factors *TWIST* and *MSX2*. Gain-of-function mutations in the *FGFR* family are associated with Crouzon, Pfeiffer, Apert, Jackson-Weiss, and Muenke syndromes.²⁰ Mutation of the FGF receptors results in increased affinity between the ligand and the receptor. The transforming growth factor- β (TGF- β) family has also been shown to have an important role in suture fusion, with increased signaling of TGF- β 1 and TGF- β 2 in multiple animal models.²¹ Bone morphogenetic protein (BMP), a member of the TGF- β family, has been cited as a source of abnormal suture formation. Warren et al²² surmised that syndromic *FGFR*-mediated craniosynostosis could be the result of abnormal downregulation of noggin (a BMP antagonist) expression. Murine studies showed that a mutation in the transcription factor *MSX2* appeared to have a dose-responsive effect on cranial suture fusion.²³ A mutation in the transcription factor *TWIST*

is responsible for Saethre-Chotzen syndrome, with in vitro studies suggesting it initiates osteoblast proliferation and differentiation.²⁴ Both MSX2 and TWIST appear to be linked to the FGF pathways. The use of transgenic mice has increased our understanding of the role of Wnt signaling in cranial suture regulation, and the use of knockouts of Axin-2 (a negative regulator of Wnt) have shown premature cranial suture fusion histologically.²⁵

Ultimately, craniosynostosis probably results from the interaction between proliferation and differentiation of osteoprogenitor cells and abnormalities in cell signaling and tissue interactions within the cranial suture.²⁶

FUNCTIONAL ISSUES

Surgeons need to determine whether patients suspected of having craniosynostosis have an isolated, single-suture event or a manifestation of an underlying syndrome. Those with single-suture, nonsyndromic craniosynostosis may have relatively straightforward issues related to possible intracranial hypertension and psychosocial concerns regarding aesthetics, whereas those with craniofacial dysostosis syndromes are some of the most complex cases for craniofacial teams to manage.

Increased Intracranial Pressure and Intracranial Anomalies

In the presence of a fused cranial suture, a rapidly growing infant brain may not have enough room for full development. This is a simplistic but useful approach to understanding and recognizing the potential for the development of increased intracranial pressure (ICP) in infants with craniosynostosis. However, in addition to craniocerebral disproportion, other factors that can be involved in the development of intracranial hypertension include abnormal venous drainage anomalies, the presence of hydrocephalus, and obstructive sleep apnea. These findings are more often seen in syndromic cases.

In classic studies, Renier et al²⁷ and Thompson et al²⁸ reported on a series of patients with craniosynostosis who underwent overnight direct ICP monitoring. They focused attention on the prevalence of intracranial hypertension in single- and multiple-suture craniosynostosis. They detected elevated ICP in up to 17% of single-suture patients and in up to 47% of multiple-suture craniosynostosis patients. The studies drew awareness to the potential detrimental effect on brain development and visual function if these problems are left untreated.

Patients diagnosed with elevated ICP can be challenging to manage. Clinical findings such as bulging fontanelles, suture diastasis and failure to thrive, headache, and vomiting may indicate intracranial hypertension. However, most children typically do not have symptoms or warning signs for a prolonged period before they are diagnosed with ICP. Radiographic findings such as a copper-beaten skull or thumbprinting on the endocranial surface are nonspecific but may corroborate funduscopic findings of papilledema (Fig. 24-6). The ventricles may be either excessively small or large, and erosions of the sella turcica may be seen. The most common objective method for diagnosing intracranial hypertension is a funduscopic examination for evidence of papilledema. Investigators at the University of Michigan²⁹ studied the effectiveness of papilledema as an indicator of raised ICP. They found that the presence of papilledema was a specific (less than 98%) indicator of elevated ICP. Sensitivity, however, was age dependent. It was 100% specific in children older than 8 years of age, but its absence in younger children was not necessarily reliable for ruling out the presence of intracranial hypertension. Given its ease of application, funduscopy continues to be a useful first step for managing all patients diagnosed with craniosynostosis. Other noninvasive methods for detecting intracranial hypertension include visual evoked potentials and optic nerve sheath ultrasound.^{30,31}

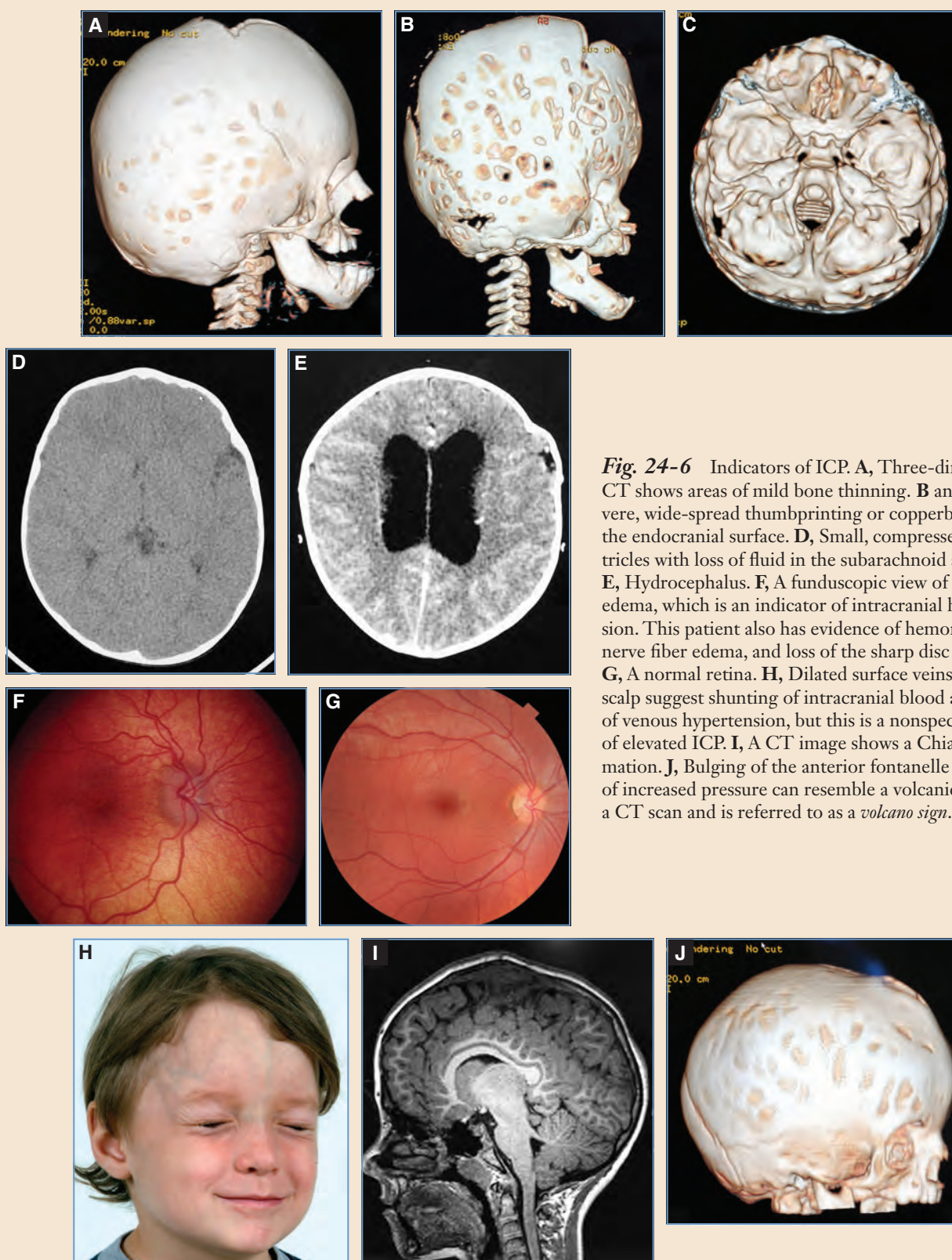


Fig. 24-6 Indicators of ICP. **A**, Three-dimensional CT shows areas of mild bone thinning. **B** and **C**, Severe, wide-spread thumbprinting or copperbeating on the endocranial surface. **D**, Small, compressed ventricles with loss of fluid in the subarachnoid space. **E**, Hydrocephalus. **F**, A fundoscopic view of papilledema, which is an indicator of intracranial hypertension. This patient also has evidence of hemorrhage, nerve fiber edema, and loss of the sharp disc margin. **G**, A normal retina. **H**, Dilated surface veins on the scalp suggest shunting of intracranial blood as a result of venous hypertension, but this is a nonspecific sign of elevated ICP. **I**, A CT image shows a Chiari malformation. **J**, Bulging of the anterior fontanelle because of increased pressure can resemble a volcanic crater in a CT scan and is referred to as a *volcano sign*.

The benchmark for assessing ICP is direct intracranial monitoring using a variety of techniques (extradural, subdural, and intraparenchymal monitors). Given its invasive nature, some centers perform this sparingly. The correlation of direct ICP measures and clinical implications is controversial. Specifically, “normal” ICP levels in nonaffected growing infants and children are not known, and adult values (higher than 20 mm Hg) may not have significance in pediatric patients. Therefore, in patients selected for direct ICP measurement, mean ICP levels with the presence of plateau waves are considered to be the most sensitive indicators of intracranial hypertension. Recent literature on the presence of ICP elevations in nonoperated, single-suture nonsyndromic craniosynostosis patients reports that up to 44% of children with sagittal and unicoronal involvement may have high ICP levels.^{32,33} However, the effect of elevated ICP on asymptomatic children is not known. Time and continued research will help to determine the clinical implications of numerically elevated pressures in clinically asymptomatic patients. ICP should be assessed in all patients with craniosynostosis.

Intracranial anomalies, hydrocephalus, and increased ICP are much more commonly seen in syndromic compared with nonsyndromic craniosynostosis patients. Intracranial hypertension has been reported in up to 50% of syndromic craniosynostosis patients, and those with Crouzon and Pfeiffer syndrome are most often affected.^{34,35} Brain anomalies in patients with syndromic craniosynostosis can be the direct result of the genetic defect or secondary to compression from craniosynostosis, raised ICP, or hydrocephalus. Brain volumes have not been found to be different in syndromic craniosynostosis patients compared with volumes in normal controls.³⁶ Venous anomalies can contribute to intracranial hypertension in children with syndromic craniosynostosis. Jugular foramen stenosis or atresia can lead to the development of collateral emissary veins, which may represent the main route for venous drainage of the cerebral venous system (Fig. 24-7). Anomalies of the venous sinuses have been identified in up to 80% of children with syndromic craniosynostosis and warrant preoperative imaging.³⁷

Hydrocephalus or ventricular dilation is present in about 40% of syndromic craniosynostosis patients.³⁸ The pathogenesis is unclear, but potential causative factors include a hypoplastic and constricted posterior fossa, venous outlet occlusion, primary brain maldevelopment, and a compensated state of increased cerebrospinal fluid outflow resistance. Crouzon and Pfeiffer syndromes are more likely to require surgical intervention in the form of a third ventriculostomy or shunt, whereas Apert syndrome is usually managed with a nonprogressive ventriculomegaly.³⁸

A Chiari malformation (herniation of the cerebellar tonsils through the foramen magnum) is frequently seen in multiple-suture and syndromic craniosynostosis patients (see Fig. 24-6, I). This should be documented and followed radiologically as part of the management of these patients. It is thought to result from a discrepancy between growth of the hindbrain and a small posterior fossa. The optimal management is controversial, but the association of increased ICP and Chiari malformation may necessitate a posterior cranial vault expansion and/or decompression of the foramen magnum.³⁹

Psychosocial Implications

In a single-suture nonsyndromic craniosynostosis, surgical intervention in a child with a craniofacial difference is indicated mainly to correct visual facial and cranial differences to improve craniofacial balance and quality of life. Significant evidence shows the negative psychosocial impact of a visible facial difference on behavior, social interaction, peer support, and psychiatric disease. Surgical improvement has a profound positive effect in many of these areas.^{40,41}

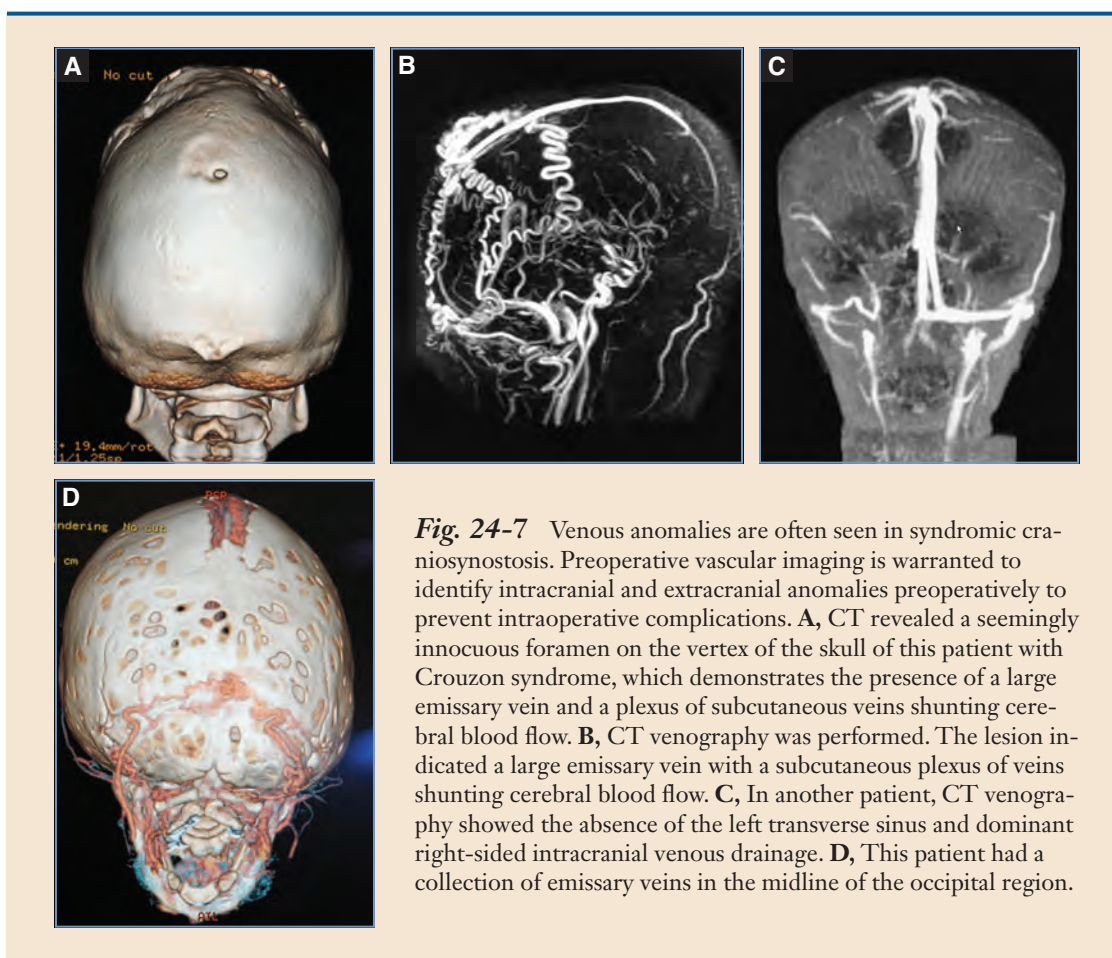


Fig. 24-7 Venous anomalies are often seen in syndromic craniosynostosis. Preoperative vascular imaging is warranted to identify intracranial and extracranial anomalies preoperatively to prevent intraoperative complications. **A**, CT revealed a seemingly innocuous foramen on the vertex of the skull of this patient with Crouzon syndrome, which demonstrates the presence of a large emissary vein and a plexus of subcutaneous veins shunting cerebral blood flow. **B**, CT venography was performed. The lesion indicated a large emissary vein with a subcutaneous plexus of veins shunting cerebral blood flow. **C**, In another patient, CT venography showed the absence of the left transverse sinus and dominant right-sided intracranial venous drainage. **D**, This patient had a collection of emissary veins in the midline of the occipital region.

Neurodevelopmental Implications

The evidence supporting an association between nonsyndromic craniosynostosis and neurodevelopmental delay is controversial. Two factors have most often been cited as causal links between single-suture craniosynostosis and impaired function: elevated ICP with hypovascularity and secondary cerebral deformation resulting from brain growth in an abnormally shaped skull.^{27,42,43} Speltz et al⁴⁴ reviewed 17 studies between 1972 and 2003, examining the neuropsychological status of children with single-suture craniosynostosis. Most children had global developmental or IQ scores within the normal range; however, 35% to 40% of assessed cases demonstrated an adverse neurodevelopmental outcome such as learning disability, language impairment, or a less precisely defined “behavioral or cognitive abnormality.” No predilection toward a specific suture was identified. In a more recent review of patients with single-suture craniosynostosis, Kapp-Simon et al⁴⁵ concluded that although global measures of intelligence and developmental status were average to low average in levels of performance, little evidence showed mental retardation or significant global deficits in cognitive function. More concerning, however, is the growing amount of evidence of school-age and older children with single-suture craniosynostosis having

deficits in neuropsychological functions, including attention and planning, processing speed, visual spatial skills, and problems in language, reading, and spelling.⁴⁵ Isolated sagittal synostosis is associated with speech and language impairment in up to 50% of affected subjects.⁴⁶

The relationship between a patient's surgical age and long-term neuropsychological outcomes in sagittal-suture craniosynostosis is equivocal. A statistically significant higher percentage of patients treated after 6 months of age had one or more reading-related learning disabilities, compared with those undergoing earlier surgery.⁴⁷ Furthermore, a comparative analysis examining the effects of two techniques related to long-term intellectual functioning showed that the type of surgical intervention for isolated sagittal synostosis may affect long-term neuropsychological outcomes. Patients undergoing early, whole-vault cranioplasty attained a higher intelligence quotient and achievement scores relative to those undergoing strip craniectomy. Surgical management with whole-vault cranioplasty performed before 6 months of age provides the most favorable long-term intellectual outcomes in patients with isolated sagittal synostosis.⁴⁸ These outcomes need to be duplicated in other centers. They show that progress needs to be made in understanding the implications of and outcomes after surgery for craniosynostosis.

Children with syndromic craniosynostosis have a higher incidence of neurodevelopmental and intellectual delay.⁴⁹ Although intracranial hypertension is often thought to be a direct cause, evidence is circumstantial. Craniocerebral disproportion may be a mechanical cause for brain anomalies such as brain distortion and cerebellar tonsillar herniation. However, it cannot be definitively linked to neurodevelopmental delay. Recent evidence demonstrates that cell adhesion molecules (L1 cell adhesion molecule) are important in the development of the white matter, requiring interactions with FGFRs.⁴⁹ *FGFR* gene mutations common to syndromic craniosynostosis may impair this interaction and lead to a variety of brain anomalies involving the white matter (for example, ventriculomegaly, callosal agenesis, agenesis of the septum pellucidum, pyramidal hypoplasia, and temporal white matter deficiencies).⁴⁹

Visual Issues

In single-suture craniosynostosis, orbital anatomy rarely affects visual function. Patients with metopic synostosis have mild hypotelorism and a pseudoesotropia resulting from the presence of epicanthal folds, but vision is usually normal. However, infants with unicoronal craniosynostosis have a high rate of amblyopia or strabismus (both 50%), but the unique shape of the plagiocephalic orbit has not proved causative.⁵⁰

The bony orbit in craniofacial dysostosis syndromes is very abnormal—often shallow and small—resulting in exorbitism and proptosis. This places the globe at risk for exposure keratitis and ulceration from lagophthalmos. Intracranial hypertension may result in irreversible optic nerve atrophy. The most common causes of visual loss are ametropia, optic atrophy, and amblyopia. Children with syndromic craniosynostosis have been found to have significant visual field deficits, as measured by kinetic visual field examination and visual evoked potentials.⁵¹ Up to 50% of children with syndromic craniosynostosis have visual impairment.⁵²

Airway Issues

Children with craniofacial dysostosis syndromes often have airway problems because of anatomic and physiologic abnormalities. They are predisposed to airway obstruction from midface retrusion caused by maxillary hypoplasia, choanal atresia, and tracheal and laryngeal anomalies. Laryngomalacia, gastroesophageal reflux, reactive airways disease, and central apnea can exacerbate these anatomic abnormalities.^{53,54} Sleep assessment has become an integral component of the workup of syndromic craniosynostosis patients. Specifically, during rapid eye movement sleep,

skeletal muscle relaxation may lead to airway obstruction with secondary increased ICP from hypercapnea-induced cerebral vasodilation. In children with preexisting compromised airways, obstructive sleep apnea will further contribute to intracranial hypertension. Airway management includes positioning measures (prone), nasopharyngeal tubes, bone distraction procedures, and in few patients, a tracheostomy. For syndromic patients who require a tracheostomy, anatomic anomalies are often identified, including a tracheal cartilaginous sleeve and a figure-of-eight trachea.⁵⁵ A thorough airway evaluation is required in infants with syndromic craniosynostosis.

PREOPERATIVE ASSESSMENT

Imaging Studies

For children with single-suture nonsyndromic craniosynostosis, a diagnosis relies heavily on the clinical assessment. Multiple-suture and syndromic craniosynostosis require a more thorough investigation, however.

Plain Radiographs

Generally, plain radiographs of the skull have limited application in the diagnosis and assessment of infants and children with craniosynostosis. The head shape, the absence of defined major sutures, elevation/sclerosis along the suture line, and evidence of thumbprinting are nonspecific findings suggestive of craniosynostosis. The presence of a harlequin-eye deformity from the vertical orientation of the orbital roof in unicoronal synostosis is one of the few signs seen on plain radiographs that is diagnostic for craniosynostosis (see Fig. 24-11). In metopic synostosis, the orbits have a trapezoidal shape that can be seen on plain films.

Computed Tomographic Scans

CT is the highest-standard modality for the radiographic evaluation of children with craniosynostosis. It is useful for definitively diagnosing craniosynostosis and for providing important information about the head shape, bony architecture, suture pattern, brain anomalies (for example, Chiari malformations, hydrocephalus, and white matter anomalies), and vascular anatomy. CT imaging and vascular anatomy assessment are necessary in syndromic craniosynostosis patients, but their application in nonsyndromic craniosynostosis cases is controversial.⁵⁶

CT in infants and children with single-suture craniosynostosis is a concern because of the negative impact of ionizing radiation exposure and because of the possibility of developing malignancy later in life. Most craniofacial surgeons agree that CT scans are not necessary for diagnosing craniosynostosis in most nonsyndromic single-suture cases, because clinical assessment is highly accurate in experienced hands.⁵⁷ Sagittal suture synostosis is an exception, because other pathology, such as prematurity and ethnic and nutritional issues, can result in a similar scaphocephalic head shape. Studies have shown that reduced exposure to radiation using low-dose protocols can produce CT images of satisfactory quality to reveal suture patency and intracranial pathology.⁵⁸ In our institution, preoperative CT scans are employed using low-dose, bone-only windows, and adhere to the “as low as reasonably achievable” principle.

Cone-beam CT is a new modality that has the same advantages of lower doses of radiation. It is widely used in dentomaxillofacial planning, with increasing application for evaluation of the craniofacial skeleton.⁵⁹

Magnetic Resonance Imaging

MRI may be useful as an adjunct, because it is a superior modality for demonstrating intracranial and brain pathology without the risk of exposure to ionizing radiation. However, infants and young children require a general anesthetic to undergo this imaging study. MRA can be useful

but may not have the same degree of resolution as CT-based studies. Prenatal MRI has been employed in the diagnosis of intrauterine craniofacial abnormalities detected by ultrasound.

Ultrasonography

High-frequency two-dimensional and three-dimensional prenatal ultrasound after 15 weeks' gestation can accurately detect suture width and patency, in addition to providing useful information about head shape, and may be used to diagnose craniosynostosis.^{60,61} Postnatal ultrasound occasionally is a useful adjunct for ruling out craniosynostosis, although it tends to be user dependent with limited application.⁶²

Diagnosis

All infants and children with a suspected diagnosis of craniosynostosis should undergo a multidisciplinary clinical assessment. Input from the following specialties is essential in creating a management plan, depending on the presence of syndromic pathology and the age of the patient: plastic surgery, neurosurgery, ophthalmology, genetics, otolaryngology, occupational therapy, social work, psychiatry, oromaxillofacial surgery, orthodontics, dentistry, and nursing. Parental and family support is a powerful adjunct through special interest groups or approved internet sites.

Establishing a diagnosis of craniosynostosis starts with an appropriate clinical assessment that includes the history of the pregnancy, a possible family history of craniosynostosis, airway and feeding issues, and the general health and well-being of the infant. The following are examined: the shape of the cranial vault, the presence of ridging, the nature of the fontanelles, the head circumference, the cranial index (length/width ratio), the size and position of the orbits, facial asymmetry, the projection of the midface, and the intraoral region. Paradoxically, examining the extremities may provide the first clue to the presence of syndromic pathology, such as the presence of complex acrocephalosyndactyly (Apert syndrome) or broad distal phalanges in the thumbs and great toes (Pfeiffer syndrome).

Single-Suture Synostosis

Sagittal Synostosis

The sagittal suture is the most-often involved suture in craniosynostosis. It affects 1 in 2500 live births and is responsible for 40% to 55% of all forms of craniosynostosis. The male/female ratio is 4:1. Familial recurrence has been described in 6% of cases.⁶³

When the sagittal suture fuses, the coronal and lambdoid sutures compensate by increasing bone deposition in the frontal and occipital bones. Meanwhile, the metopic suture compensates with symmetrical bone expansion. This compensatory growth leads to a boat-shaped cranial vault, known as *scaphocephaly*, with varying degrees of frontal and occipital bossing. The forehead is high and prominent. The AP length of the skull increases, with transverse narrowing in the biparietal and bitemporal region (Fig. 24-8). Occasionally, the midvault region has a saddle-shaped deformity. This results in a lower than normal cranial index (width/length ratio).

CT features are consistent with clinical findings for head shape. The sagittal suture may have varying degrees of partial fusion (anterior and posterior), but this does not correlate well with the severity of the deformity.

Metopic Synostosis

The metopic suture is the first suture closed, between 3 and 8 months of age, and is the only cranial suture to fuse in childhood.⁶⁴ The incidence of metopic synostosis was thought to be 1 in 15,000 but may be closer to 1 in 10,000 with newer epidemiological data. Metopic synostosis previously was considered relatively uncommon, following unilateral coronal synostosis (the sec-

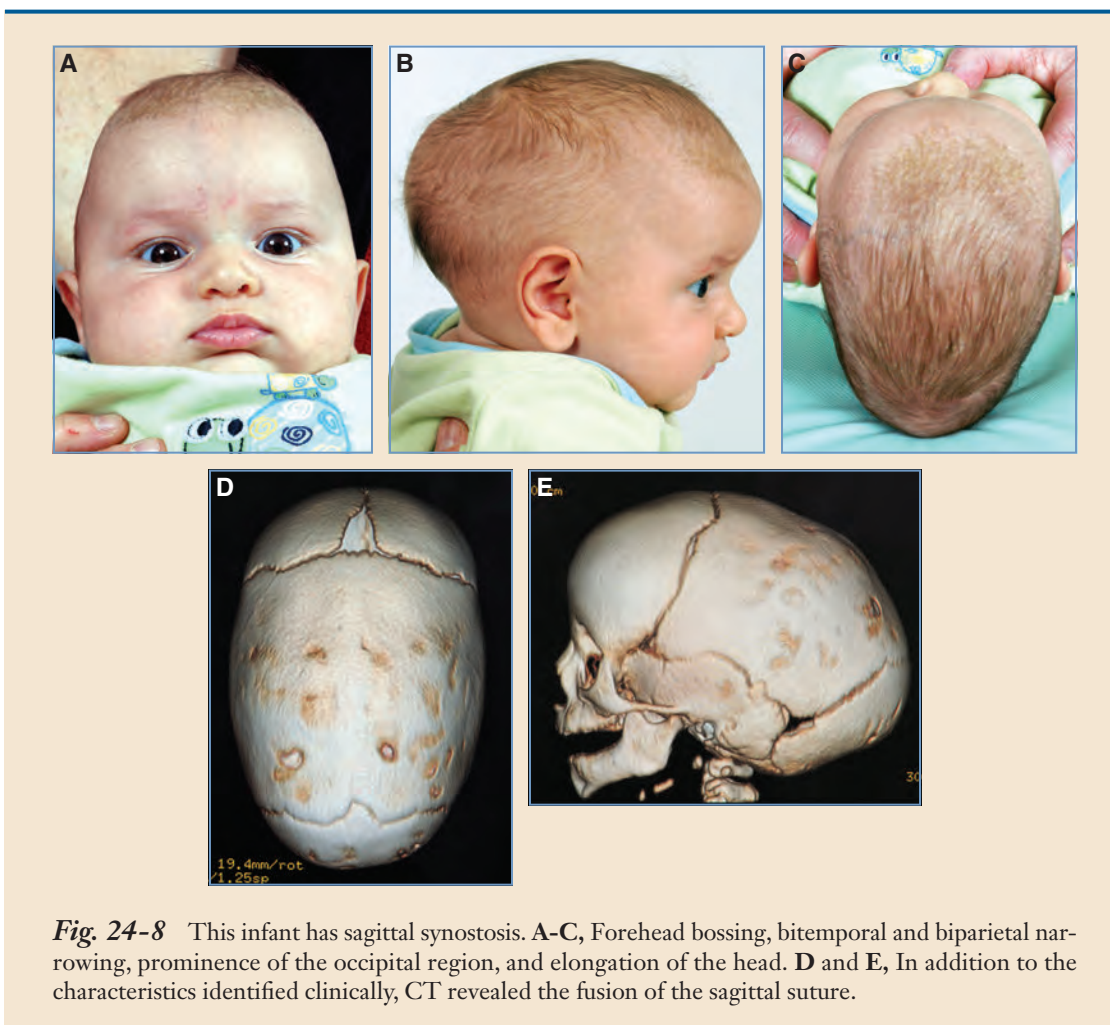


Fig. 24-8 This infant has sagittal synostosis. **A-C**, Forehead bossing, bitemporal and biparietal narrowing, prominence of the occipital region, and elongation of the head. **D** and **E**, In addition to the characteristics identified clinically, CT revealed the fusion of the sagittal suture.

and most common type) in incidence.^{65,66} However, craniofacial centers have reported that the incidence of metopic synostosis has shifted and has displaced unicoronal craniosynostosis as the second most common form seen.¹⁰ Similar to sagittal synostosis, it seems to affect more males than females, with a ratio of 3:1.⁶⁶

Premature closure of the metopic suture results in a growth restriction across the forehead and a compensatory expansion of the cranial vault posteriorly, leading to a triangle or keel-shaped forehead called *trigonocephaly* (Fig. 24-9). Additional findings include hypotelorism, pseudoesotropia, flattening of the lateral portion of the superior orbital rim, and bitemporal narrowing. Few patients will present with mild ridging along the length of the metopic suture, with no other clinical findings of trigonocephaly. This is known as *metopic ridging*. Whether this is a functional phenomenon occurring as the metopic suture normally fuses or whether it represents a form fruste of true metopic synostosis is controversial (Fig. 24-10). It is a source of concern for parents, especially when the infant presents after 8 to 10 months of age, because a CT investigation will demonstrate a fused metopic suture that can be misconstrued as a true craniosynostosis.⁶⁷ Metopic ridging rarely requires surgical attention.

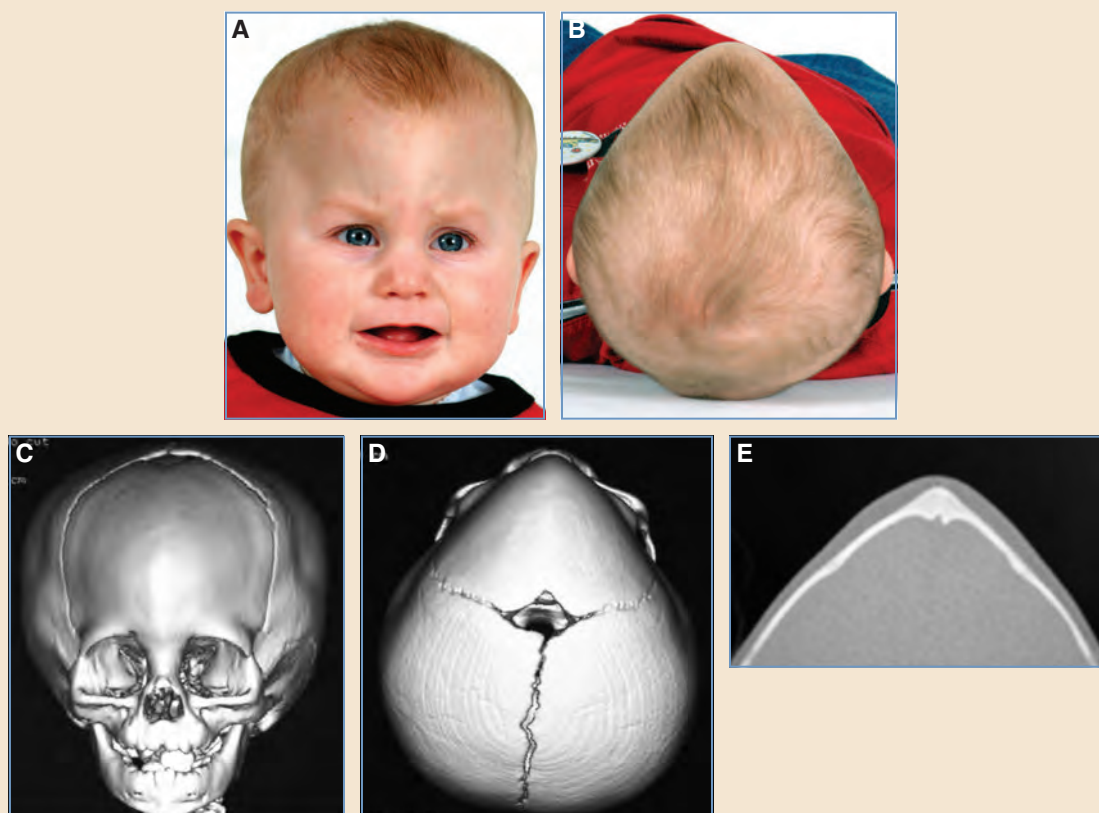


Fig. 24-9 This 8-month-old male has metopic synostosis. **A** and **B**, Hypotelorism and a midline forehead prominence were evident clinically. **C-E**, CT revealed the additional findings of a trapezoidal orbital configuration, trigonocephaly with posterior cranial vault compensatory expansion, and a small midline notch on the endocranial surface, known as an *omega sign*, often seen in metopic synostosis.

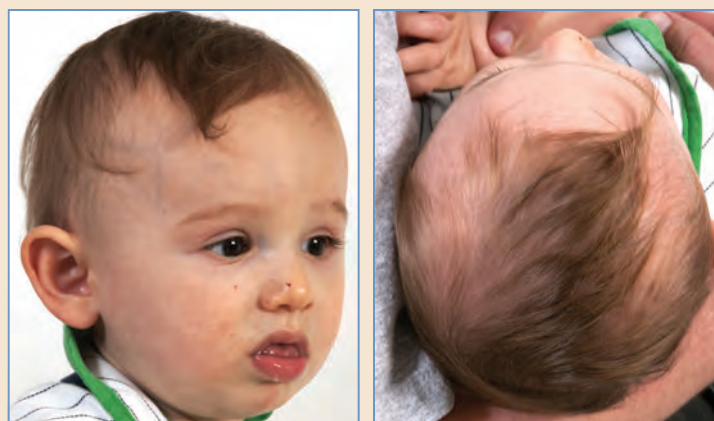


Fig. 24-10 Metopic ridging in a 10-month-old male. This may be a form fruste of metopic synostosis. Patients present with a midline ridge along the fused metopic suture but do not have the accompanying features of true metopic synostosis such as lateral orbital wall recession and hypotelorism.

More than any other single-suture craniosynostosis, trigonocephaly has been associated with syndromes such as Saethre-Chotzen, Opitz C trigonocephaly syndrome, Say-Meyer trigonocephaly syndrome, Christian syndrome, and Floating-Harbor syndrome. It has also been associated with several chromosomal anomalies such as Jacobsen syndrome.⁶⁸

CT features include a triangular shape to the head, a fused metopic suture, hypotelorism, and a characteristic trapezoidal shape to the orbits, with converging axes. One finding described to be pathognomonic of metopic synostosis is the omega sign, which is a bony groove on the endocranial surface along the length of the anterior sagittal sinus⁶⁸ (see Fig. 24-9).

Unilateral Coronal Synostosis

Unilateral coronal synostosis results in asymmetrical bossing of the forehead, which is *anterior plagiocephaly*. It was reported as the second most common type of craniosynostosis (20% to 25%), occurring in 1 in 15,000 live births; however, recent data trends suggest that metopic synostosis occurs more frequently than unilateral coronal synostosis.⁹ Unicoronal synostosis is more common in females and more common on the left side. Molecular genetic screening is recommended to determine the presence of *FGFR3* mutation associated Muenke syndrome. Visual anomalies such as strabismus and amblyopia affect up to 50% of patients with unicoronal synostosis.⁵⁰

Although unicoronal synostosis involves the fusion of only one suture, it has a characteristic bilateral deformity. Patients have forehead flattening and bulging in the temporal region on the affected side and contralateral forehead bossing from compensatory growth. The nasal root is deviated and points to the side of the synostosed suture (Fig. 24-11). Orbital asymmetry is obvious; the orbit on the affected side is larger and rounder than the smaller, beady-eyed orbit on the contralateral side.

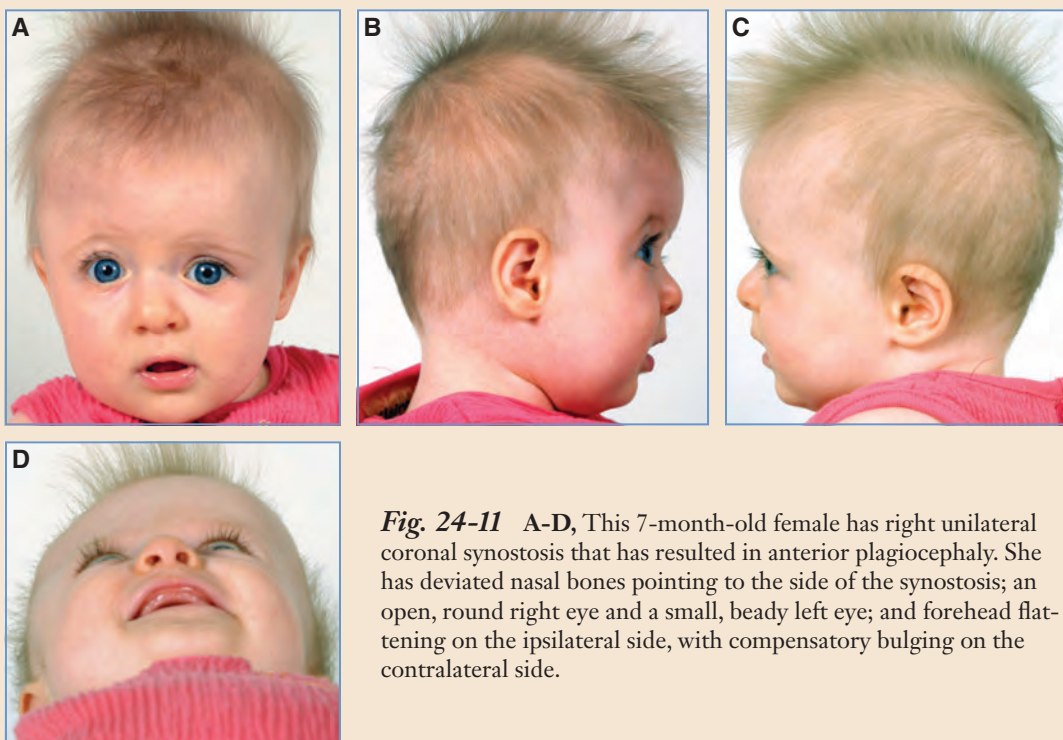


Fig. 24-11 A-D, This 7-month-old female has right unilateral coronal synostosis that has resulted in anterior plagiocephaly. She has deviated nasal bones pointing to the side of the synostosis; an open, round right eye and a small, beady left eye; and forehead flattening on the ipsilateral side, with compensatory bulging on the contralateral side.

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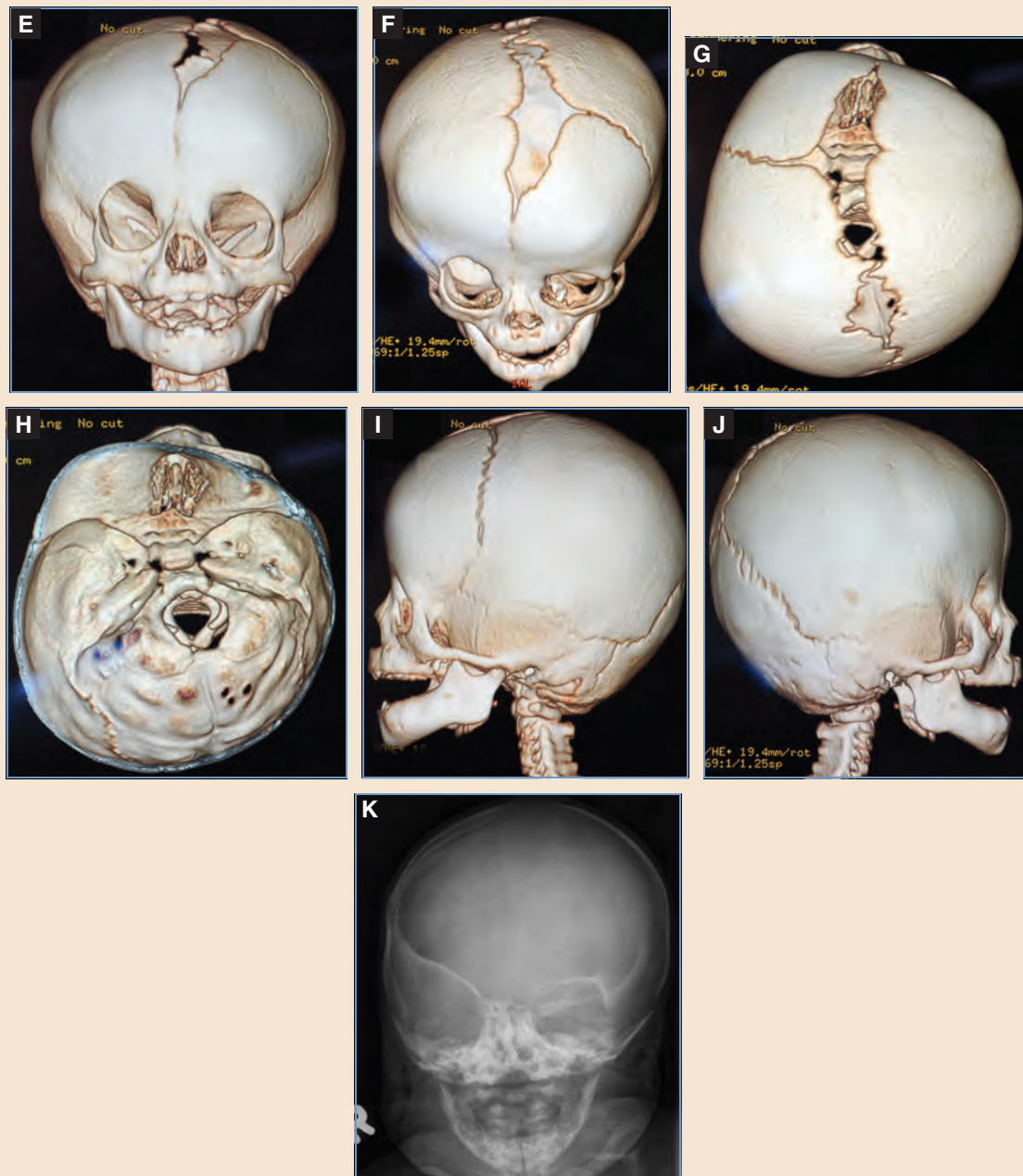


Fig. 24-11, cont'd E-J, CT images show posterior angulation of the lateral orbital wall on the side of the fused suture and significant cranial base asymmetry. K, A plain radiograph demonstrates vertical orientation of the sphenoid wing, known as a *harlequin-eye sign*, on the side of the synostosed coronal suture.

CT findings include foreshortening of the anterior cranial fossa on the side of the synostosed suture, a harlequin-eye deformity from ventral displacement of the greater wing of the sphenoid in the orbital roof, an acute angulation between the lateral orbital wall and the zygomatic arch, and deviation of the anterior fontanelle to the side opposite the affected suture.

Bilateral Coronal Synostosis

Isolated nonsyndromic bilateral coronal synostosis is quite rare and more frequently seen in the myriad craniosynostosis syndromes. The presence of midface retrusion may be an indicator of syndromic pathology. The coronal ring consists of the paired coronal sutures and the fronto-sphenoidal and sphenothmoidal sutures along the anterior cranial base. Fusion of these cranial sutures restricts AP growth of the cranium, resulting in brachycephaly. Compensatory growth through the sagittal and squamosal sutures causes vertical elongation of the skull (turriccephaly), and often the two deformities coexist (turribrachycephaly). Bilateral temporal bulging occurs frequently, with recession of the supraorbital rim behind the anterior projection of the cornea (Fig. 24-12). CT findings include bilateral harlequin-eye deformities and severe shortening of the anterior cranial fossa. The cranial base has a transverse oval shape. Delayed onset of bilateral coronal sutures has been described as *oxycephaly*.

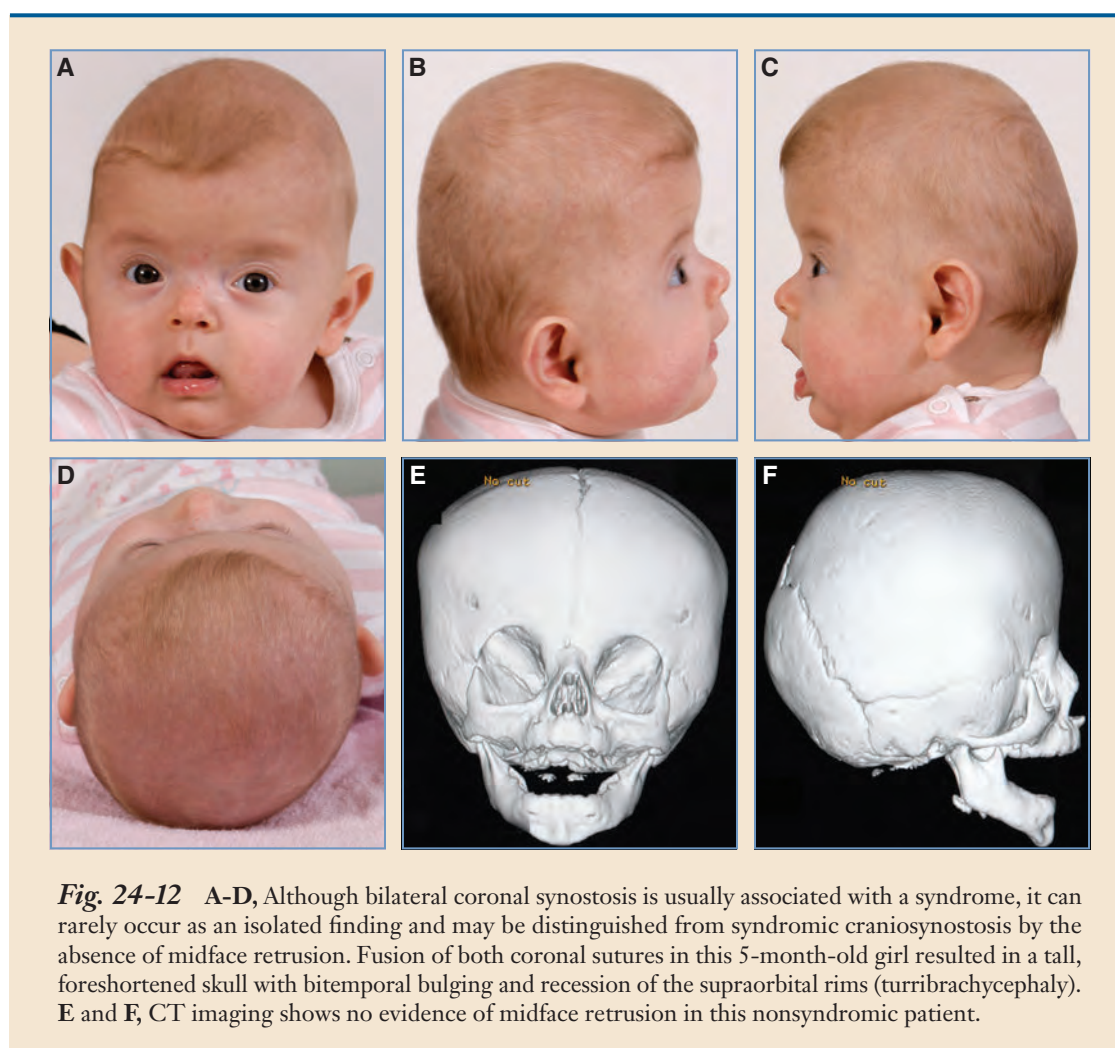


Fig. 24-12 A-D, Although bilateral coronal synostosis is usually associated with a syndrome, it can rarely occur as an isolated finding and may be distinguished from syndromic craniosynostosis by the absence of midface retrusion. Fusion of both coronal sutures in this 5-month-old girl resulted in a tall, foreshortened skull with bitemporal bulging and recession of the supraorbital rims (turribrachycephaly). E and F, CT imaging shows no evidence of midface retrusion in this nonsyndromic patient.

Lambdoid Synostosis

Of the major cranial sutures, lambdoid synostosis is the rarest form of premature suture fusion, comprising 1% to 5% of all craniosynostoses and occurring in 1 in 100,000 live births.^{69,70} It is difficult to diagnose clinically but is classically characterized by ipsilateral occipital flattening, posteroinferior displacement of the ipsilateral ear, bossing of the ipsilateral mastoid, and ipsilateral decreased height of the cranial vertex (Fig. 24-13). These findings combine to form a trapezoidal skull shape when viewed from above. From behind, cranial base canting and inferior displacement of the ipsilateral ear are seen.

Lambdoid synostosis needs to be distinguished from deformational plagiocephaly (plagiocephaly without synostosis, known as *positional plagiocephaly*), which has become exceptionally common because of prone sleeping practices designed to reduce the incidence of sudden infant death syndrome.⁶⁹ Deformational plagiocephaly has a characteristic parallelogram shape when viewed from above (Fig. 24-14 and Table 24-3). Specifically, patients have occipital flattening associated with anterior positioning of the ipsilateral ear and forehead. The ear position is equal

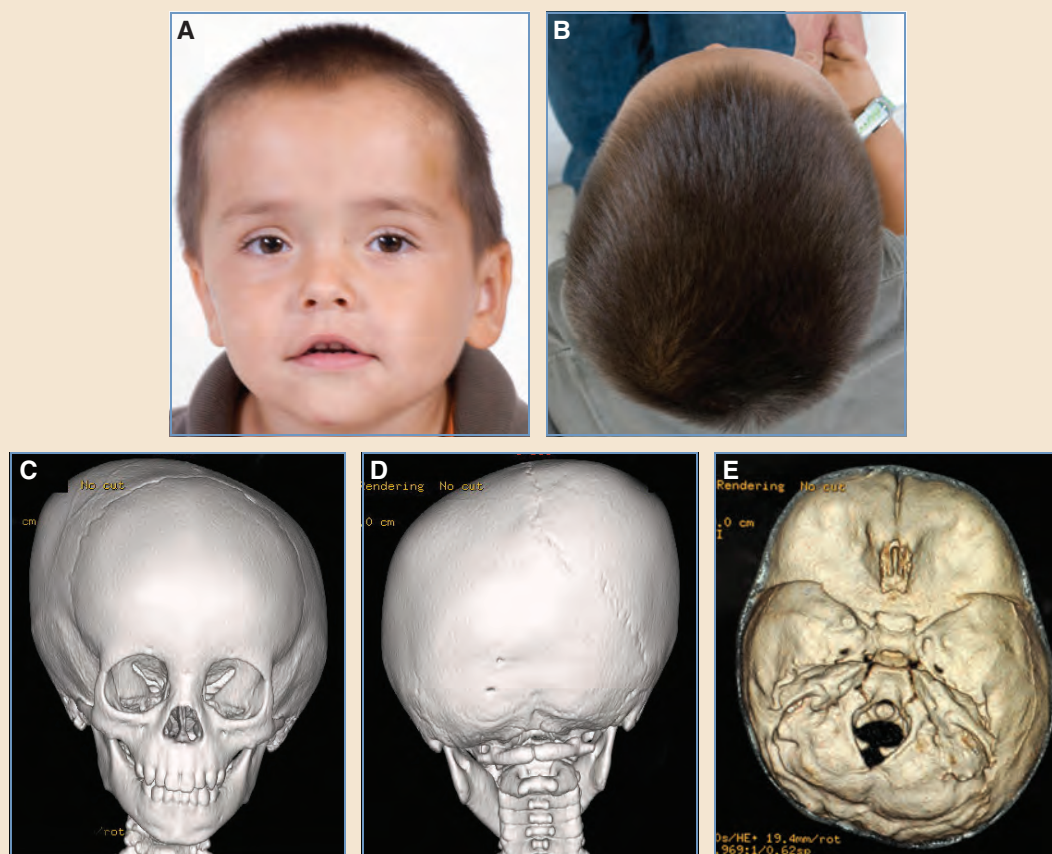


Fig. 24-13 A and B, Left lambdoid synostosis in a young boy. There is asymmetry present in the frontal view and a trapezoidal shape to his head from above. C-E, CTs demonstrate left lambdoid suture fusion with a compensatory vertical prominence on the opposite side of the skull; canting of the skull base on the posterior view with a mastoid bulge on the side of the synostosed suture; and marked asymmetry and volumetric differences between the two sides of the posterior fossa on the cranial base view.

when viewed from behind no matter how severe the deformity. The incidence of deformational plagiocephaly is very common, affecting up to 1 in 7 infants. Many of the babies are born with round heads, whereas those with lambdoid synostosis have the deformity at birth.⁷⁰

CT findings demonstrate the deformities described previously and significant asymmetry in the cranial base (see Fig. 24-13).

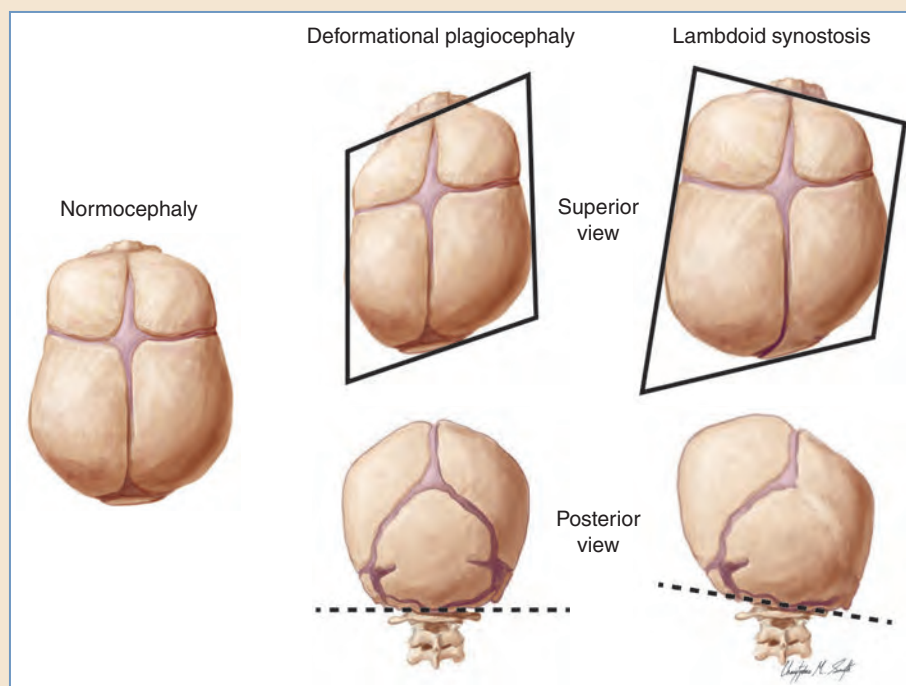


Fig. 24-14 Differentiation of deformational plagiocephaly and lambdoid synostosis.

Table 24-3 Comparison of Plagiocephaly and Lambdoid Synostosis

	Positional Plagiocephaly	Lambdoid Synostosis
Age at onset	2+ weeks after birth	Present at birth
Bird's-eye view of head shape	Parallelogram	Trapezoidal
Forehead on the affected side	Anteriorly displaced	Symmetrical or retruded
Ear position on the affected side	Anteriorly displaced	Posteriorly displaced
Skull base viewed from behind	Ears level	Ears uneven (the affected side is lower)
Natural history	Improvement*	Worsens during the first year of life, then stabilizes
Incidence	Very common	Very rare (from 1 in 100,000 to 1 in 130,000) ⁷⁰

*Assuming the patient has no torticollis or significant developmental delay.

Multiple-Suture Synostosis

Approximately 5% of all cases of craniosynostosis involve multiple sutures.⁷¹ Almost any pattern of multiple-suture synostosis can exist, but involvement of both lambdoid and sagittal sutures has been formally recognized as the *Mercedes-Benz* pattern of craniosynostosis, referring to the three-pointed star emblem of the automobile company.^{72,73} Patients with multiple-suture involvement have complex problems and often have associated Chiari malformations and signs of intracranial hypertension.

Minor Suture Variants

Although fusion of the minor sutures is less common, it can alter the skull shape and make diagnosis challenging. Isolated frontosphenoidal synostosis may mimic unicoronal synostosis and produce anterior plagiocephaly.⁷⁴ Frontozygomatic suture synostosis has been reported.^{75,76} Distinguishing these variants will rely on CT imaging.

Syndromic Craniosynostosis

A syndrome can be considered a constellation of signs and symptoms that, together, characterize a clinical condition. Craniosynostosis has been detected in more than 180 different syndromes, and the number continues to increase as sophisticated molecular genetic analyses help to identify new syndromes.⁷¹ Identifying syndromic craniosynostosis requires a combination of clinical, radiologic, and molecular genetic assessment. Phenotype alone may not be a reliable indicator of the type of syndrome, because several may share similar craniofacial suture involvement, often including the coronal ring (see Fig. 24-12). The main craniosynostosis syndromes are discussed in the following sections.

Apert Syndrome

Apert syndrome was first described in 1894 and is characterized by bilateral coronal craniosynostosis, midface retrusion, exorbitism, complex symmetrical acrocephalosyndactyly, radiohumeral

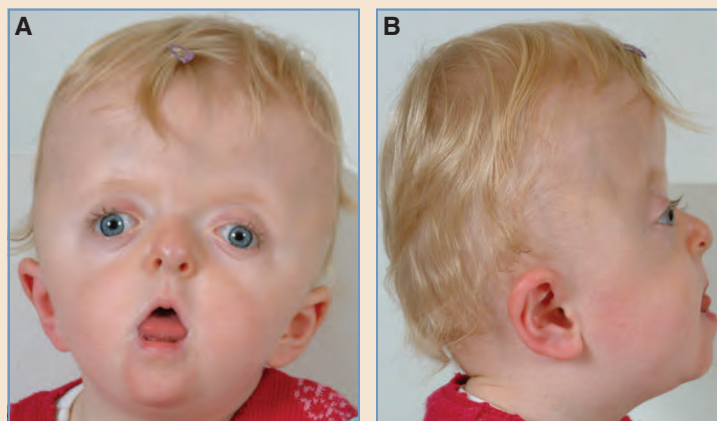


Fig. 24-15 A and B, This 11-month-old female has Apert syndrome. She has hypertelorism without severe exorbitism, turribrachycephaly from bicoronal synostosis, midface retrusion, and a parrot-beak nasal deformity.

fusion, and variable degrees of neurodevelopmental delay.⁷⁷ The nose is short and wide with a bulbous tip, and the anterior facial height is reduced (Fig. 24-15). The incidence is 1 in 70,000.⁷⁸ Apert syndrome is transmitted in an autosomal dominant fashion, although the most cases result from a spontaneous mutation in *FGFR2*. It accounts for approximately 4% of all cases of craniosynostosis.⁷⁸ The male/female ratio is equal.

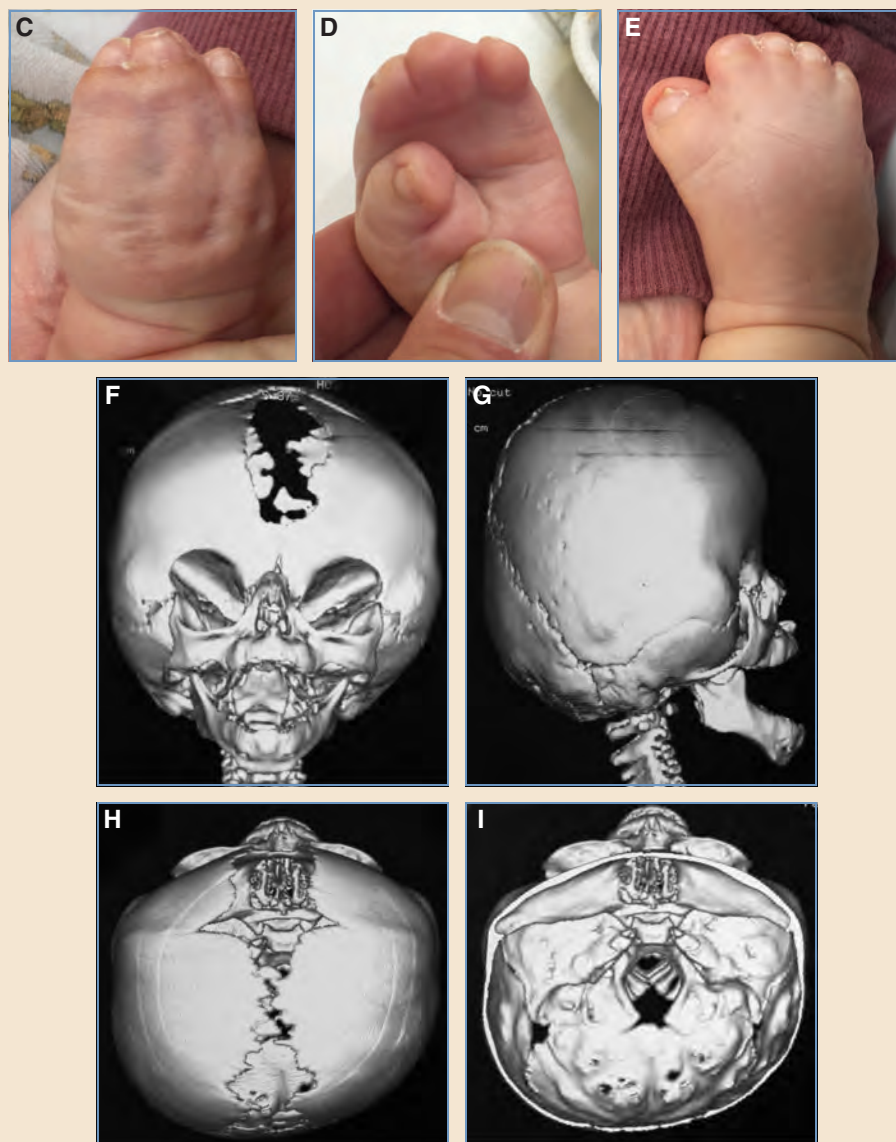


Fig. 24-15, cont'd C-E, The diagnosis is established by examining the extremities, which have complex acrocephalosyndactyly. F-I, CT images demonstrate bicoronal synostosis; a large, confluent anterior fontanelle; bitemporal bulging; divergent orbital axes; and maxillary retrusion.

Crouzon Syndrome

Crouzon first described this syndrome in 1912.⁷⁹ Clinical characteristics include bilateral coronal craniosynostosis, exorbitism with hypertelorism, maxillary hypoplasia with relative mandibular prognathism, and normal intelligence (Fig. 24-16). Delayed-onset pansynostosis with intracranial hypertension has been described in a subset of Crouzon syndrome patients and should be carefully monitored. The incidence is 1 in 65,000 live births, and the syndrome is inherited in an autosomal dominant pattern with variable penetration.⁷¹ It results from a mutation in *FGFR2*. Parental involvement is typically seen. A variant of Crouzon syndrome involves acanthosis nigricans and is associated with *FGFR3* mutations. Generally, craniofacial morphology is less severe than in Apert syndrome.⁸⁰

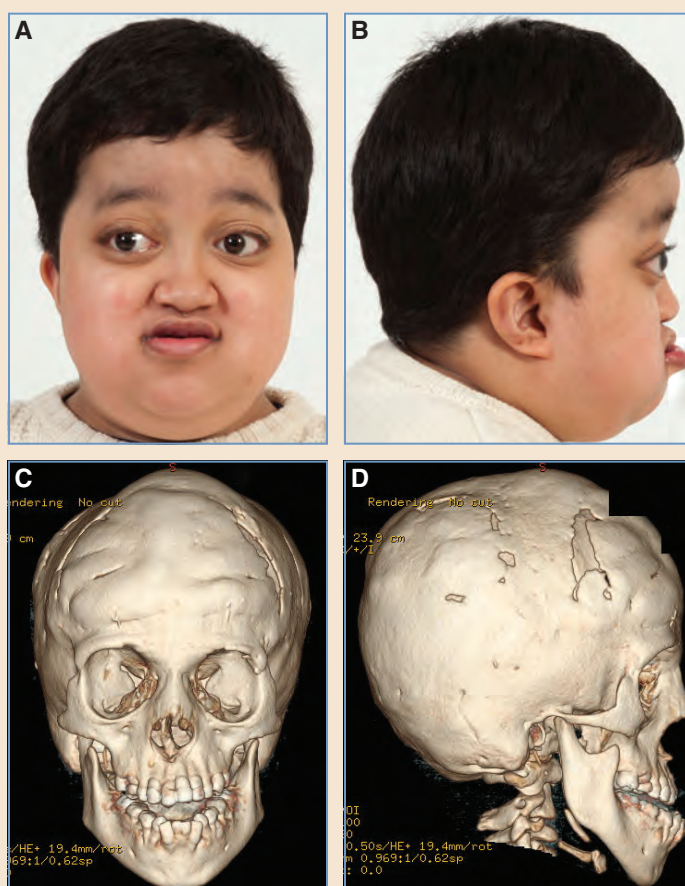


Fig. 24-16 A and B, This 8-year-old boy has Crouzon syndrome, with severe exorbitism, midface retrusion, and mild turribrachycephaly from bicoronal synostosis. C and D, CT images reveal evidence of previous cranioorbital reshaping.

Pfeiffer Syndrome

Pfeiffer syndrome presents with bicoronal synostosis and may resemble Crouzon syndrome because of the presence of proptosis, hypertelorism, and maxillary hypoplasia (Fig. 24-17). It is distinguished by extremity anomalies, namely broad great toes and thumbs, partial syndactyly, and brachymesophalangy.⁸¹ The incidence is 1 in 100,000 live births and may be transmitted in an autosomal dominant fashion. Molecular genetic analyses have identified a sporadic mutation in *FGFR1* or *FGFR2*. A new classification system stratifies patients according to functional assessment of respiratory, ocular, otologic, and neurologic status.⁸²



Fig. 24-17 A and B, This girl with Pfeiffer syndrome has severe exorbitism and midface retrusion. She has a tracheostomy for airway protection. C-E, CT shows bicoronal synostosis with midface retrusion and shallow orbits. F and G, Phenotypically, it is hard to differentiate Crouzon syndrome and Pfeiffer syndrome on the basis of facial dysmorphism. An examination of the extremities may reveal wide distal thumbs and great toes and simple syndactyly in patients with Pfeiffer syndrome.

Saethre-Chotzen Syndrome

Saethre-Chotzen syndrome is characterized by unilateral or bilateral coronal craniosynostosis, a low hairline, ptosis of the upper lids, syndactyly of the second and third digits, ear anomalies (prominent superior crus), and normal intelligence⁸³ (Fig. 24-18). The incidence is 1 in 25,000 to 1 in 50,000 births, and it is inherited in an autosomal dominant fashion and results from a mutation in *TWIST1*.⁸³ In contradistinction to other cases of syndromic craniosynostosis, midface retrusion is unusual.

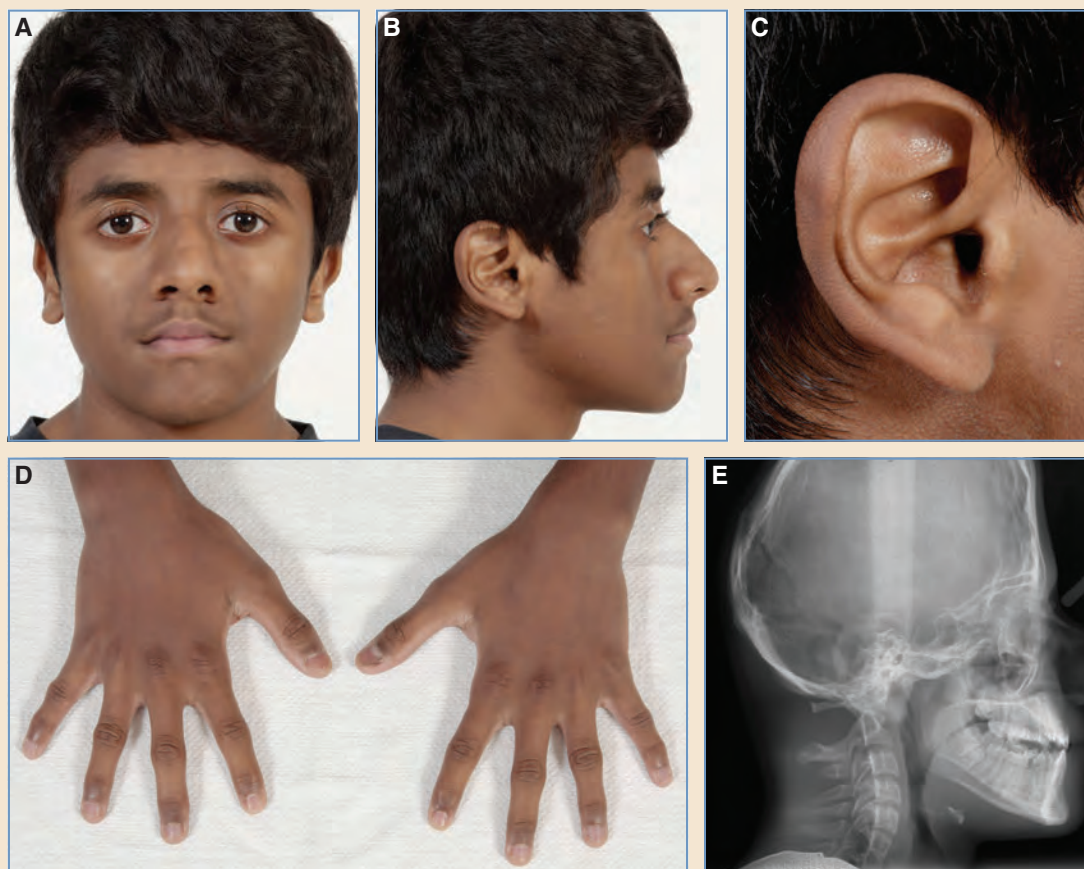


Fig. 24-18 A and B, This 13-year-old boy has Saethre-Chotzen syndrome, characterized by mild upper lid ptosis, turribrachycephaly from bicoronal synostosis, and supraorbital rim retrusion. C, His ears have characteristic prominent superior crura. D, There is evidence of bilateral simple syndactyly of the fingers. E, A lateral cephalometric radiograph shows a tall, retruded forehead; fusion of both coronal sutures; and a class I dental and skeletal relationship.

Muenke Syndrome

Patients with Muenke syndrome have a specific *FGFR3* pathogenic variant that results in the protein change Pro250Arg. The reported incidence is 1 in 30,000 live births.⁸⁴ The phenotype is highly variable and may include unilateral or bilateral coronal craniosynostosis, brachydactyly, carpal and tarsal fusions, sensorineural hearing loss, a Klippel-Feil cervical anomaly, and variable degrees of mental impairment^{84,85} (Fig. 24-19). The incidence is variable. Muenke syndrome affects up to 20% of coronal synostosis patients and is inherited in an autosomal dominant manner. Females with the Pro250Arg mutation are significantly more likely to have craniosynostosis than males.⁸⁶

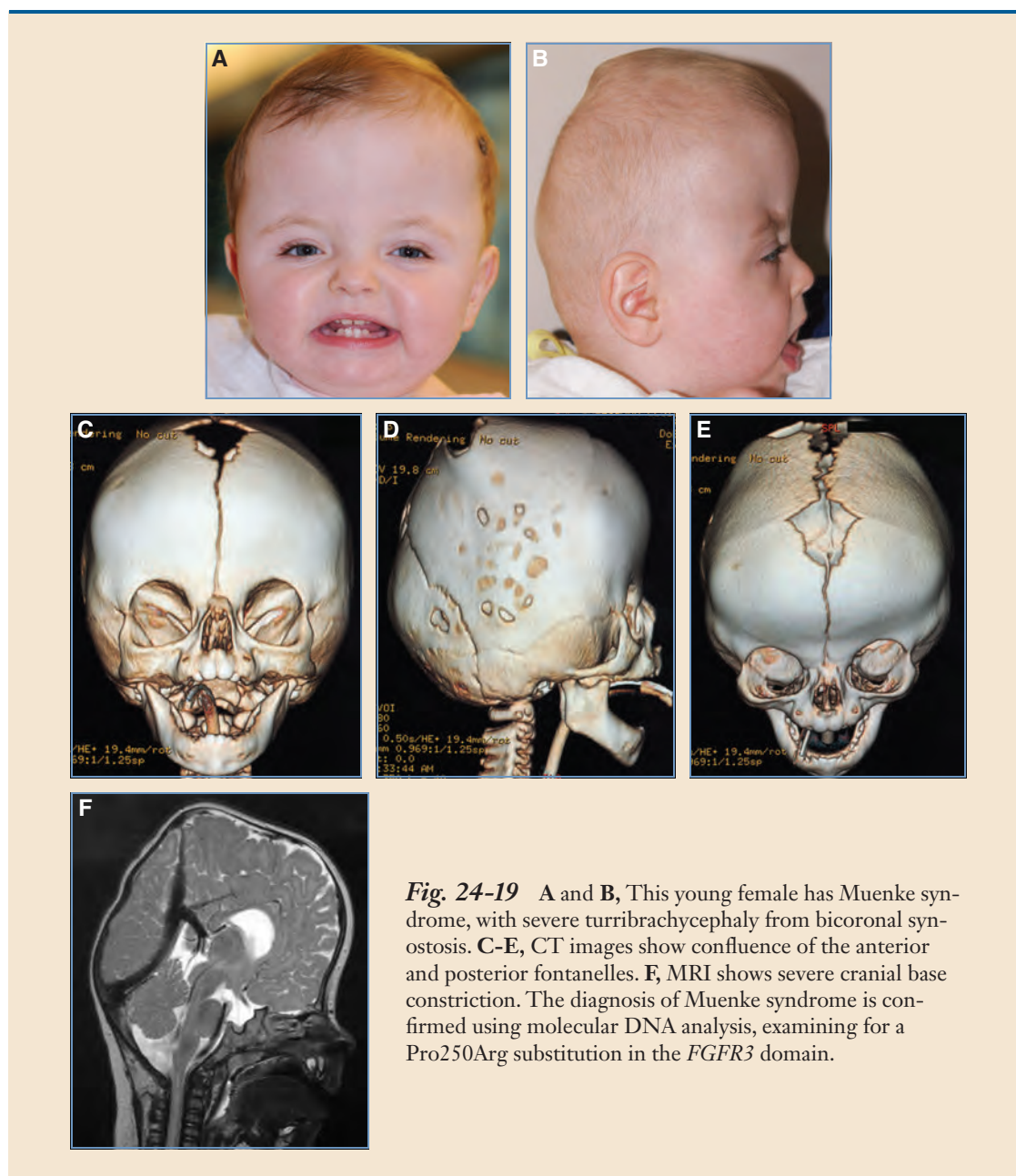


Fig. 24-19 A and B, This young female has Muenke syndrome, with severe turribrachycephaly from bicoronal synostosis. C-E, CT images show confluence of the anterior and posterior fontanelles. F, MRI shows severe cranial base constriction. The diagnosis of Muenke syndrome is confirmed using molecular DNA analysis, examining for a Pro250Arg substitution in the *FGFR3* domain.

Kleeblattschädel

Kleeblattschädel is a German word for *cloverleaf* and describes the most severe form of multiple-suture craniosynostosis. It is a clinical finding and not a diagnosis (Fig. 24-20). Synostosis usually involves the lambdoid, coronal, and skull base sutures; the sagittal and temporal squamosal sutures fuse in a delayed fashion. The degree of severity will vary according to the involvement of different sutures. With growth, compensatory bulging results in the classic trilobar or three-part cloverleaf shape of the cranial vault. The cause is heterogeneous. Kleeblattschädel often represents a manifestation of Pfeiffer or Crouzon syndrome. Intracranial pathology such as Chiari malformation and hydrocephalus is common. Significant morbidity and mortality are common.⁸⁷

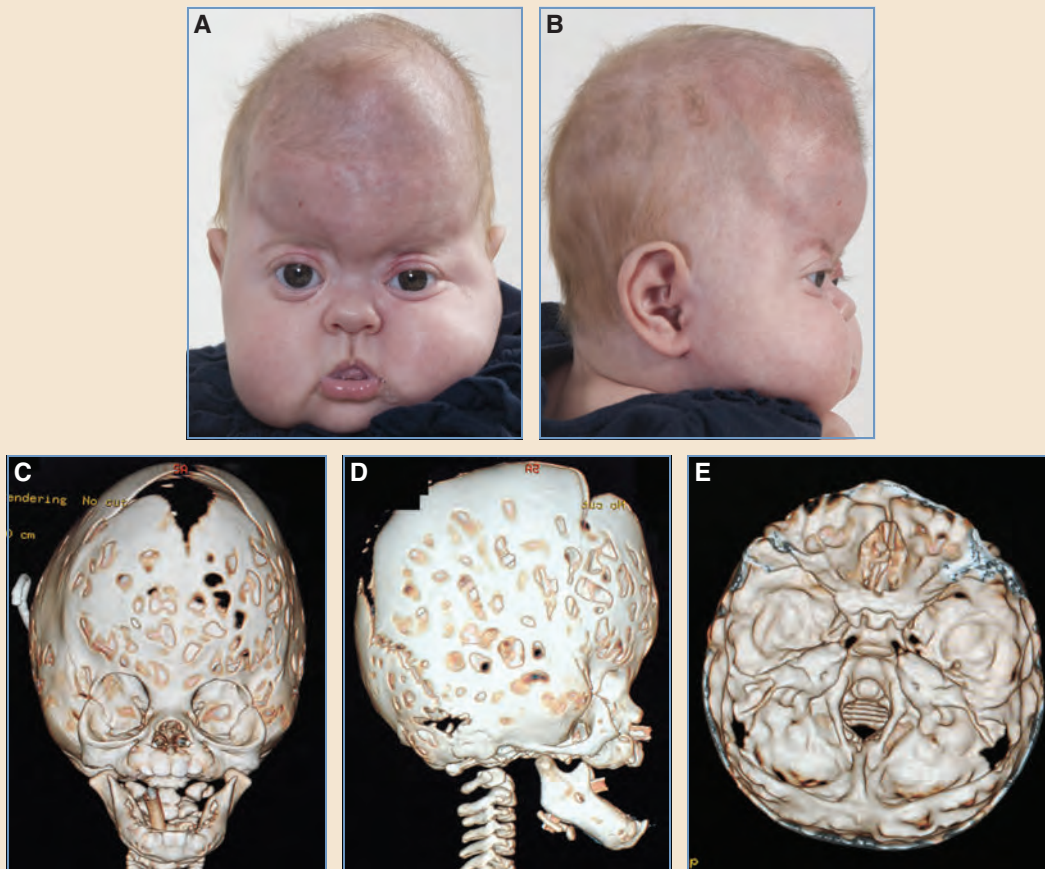


Fig. 24-20 A and B, This infant has Crouzon syndrome, with severe exorbitism, a bulging forehead, and bilateral temporal bulges. C-E, CT scans show severe thumbprinting, bicoronal synostosis with temporal squamosal and metopic involvement, and midface retrusion.

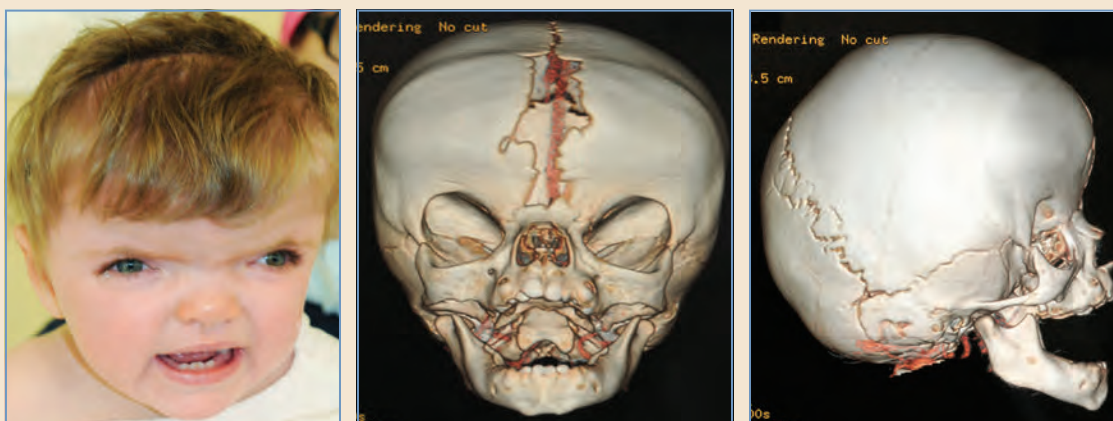


Fig. 24-21 This 8-month-old female has craniofrontonasal dysplasia, with turribrachycephaly and hypertelorism. It should be suspected in patients who have unicoronal or bicoronal synostosis and hypertelorism without evidence of midface retrusion.

Craniofrontonasal Dysplasia

Craniofrontonasal dysplasia is a rare, X-linked craniosynostosis syndrome caused by a mutation in the *EFNB1* gene.⁸⁸ It has a variable phenotypic expression, but females are more often and more severely affected than males. It commonly involves the coronal suture or sutures and is associated with orbital hypertelorism; a bifid nose; dry, frizzy hair; longitudinal ridging of the nails; and anomalies of the upper extremity (Fig. 24-21). It may be clinically distinguished from frontonasal dysplasia (which is a clefting sequence) by the presence of craniosynostosis.⁸⁸

SURGICAL APPROACHES TO CRANIOSYNOSTOSIS

Surgical correction of single-suture synostosis usually requires a single, major procedure in the first year of life. Most patients undergo significant improvement but are followed closely for the possibility of relapse and the need for a touch-up surgery. On the contrary, syndromic craniosynostosis requires a series of staged, age-dependent procedures performed from infancy to skeletal maturity. Relapse rates in patients with syndromic craniosynostosis are high, and the outcome is based more on the biology of the underlying condition than on the surgical technique. Protocols vary from center to center but essentially focus on airway compromise, cranial dysmorphism, and ocular and intracranial pathology in the first few years of life. The makeup of the surgical craniofacial team varies from center to center, but key members are from the following departments: pediatric anesthesia, nursing, plastic and reconstructive surgery, neurosurgery, oromaxillofacial surgery, otolaryngology–head and neck surgery, critical care, and ophthalmology.

Craniosynostosis surgery is designed to address cranial dysmorphology, craniocerebral disproportion, orbital position, and intracranial hypertension. The myriad techniques that have been described can be classified as static or dynamic and compressive or decompressive. Craniofacial units disagree about which is the best technique. Postoperative care is provided in critical care or high-acuity observation rooms.

History of Surgical Treatment

The surgical treatment of craniosynostosis has a long history that started in the late nineteenth century. Almost simultaneously in the 1890s, a French surgeon, Lannelongue⁸⁹ and an American, Lane,⁹⁰ described linear craniectomies and removal of the pathologic suture in an attempt to correct mental “imbecility” from premature sutural closure and microcephaly.

Despite best intentions, the mortality rate was almost 50% in the first 33 children treated in this manner. Jacobi,⁹¹ thought to be the father of American pediatrics, condemned the procedure in 1894. Interest in surgery for craniosynostosis was minimal for almost the next three decades.

Surgical techniques and anesthesia improved, and in the 1920s Faber and Towne⁹² pioneered the concept of prophylactic linear strip synostectomy. They reported excellent preservation of function with minimal morbidity and mortality using this procedure, which they recommended for infants 1 to 3 months of age.

The concept of unlocking the growth potential of the craniofacial skeleton by releasing the fused suture in the growing skull continues to resonate in the modern-day philosophy of craniosynostosis surgery. However, surgeons continued to be vexed by postoperative, rapid reossification and relapse of the deformity. This led to numerous strategies to prevent bone ingrowth, including the use of caustic solutions on the dura and the placement of silastic and metal interpositional devices.

In 1967 Tessier⁹³ introduced new techniques and principles of cranial vault remodeling, bone grafting, and stabilization. These operations were significantly more radical in their scope and correction. Tessier showed that the craniofacial skeleton could be osteotomized and reassembled, and with stabilization of the bone, this technique would lead to predictable and reproducible outcomes. He achieved these results before the advent of sophisticated CT imaging techniques, making his intuitive skills and clinical insights even more remarkable. These principles and techniques are the cornerstone of current management philosophies for craniosynostosis repair. Many of Tessier's fellows and disciples have refined his original techniques. Subsequently, sophisticated anesthesia monitoring systems, blood conservation techniques, and small titanium and bioresorbable bone fixation and bone distraction systems affected craniofacial surgery in major ways, improving safety and outcomes.

Anterior Cranioorbital Reshaping With Frontoorbital Advancement

Indications

- Anterior plagiocephaly from unicoronal synostosis or minor suture variant
- Brachycephaly from bicoronal synostosis
- Trigonocephaly from metopic synostosis
- Supraorbital rim recession or asymmetry

Procedure

Anterior cranioorbital reshaping and frontoorbital advancement is a powerful tool in craniofacial surgeons' armamentarium for correcting congenital deformities of the face and skull. This procedure removes and reshapes the bones of the forehead, supraorbital rims, and often, the temporal bone. The timing of surgery varies by center. However, for single-suture nonsyndromic synostosis, surgery usually is performed when patients are 8 to 12 months of age, when the bone is thick enough to be stabilized using plate fixation and the infant is able to withstand the rigors of anesthesia, complex surgery, and blood loss. Another advantage of surgery at this age is the reossification of bone defects that results from expansion of the craniofacial skeleton. This opportunity is lost after approximately 14 to 18 months of age as the dura loses its osteogenic potential.

Access is obtained through a bicoronal incision with subperiosteal exposure of the frontal and orbital regions. After a bifrontal craniotomy, the supraorbital rim (bandeau) is removed, reshaped, and positioned accordingly. The bone segments are fixated using bioresorbable fixation designed to resorb over a 12- to 18-month period. Alternatively, wire or suture fixation is often employed. The use of titanium plate fixation is discouraged, because passive intracranial migration occurs as the cranial vault continues to grow and may be problematic for additional surgery or trauma. The use of age-matched surgical templates has significantly improved craniofacial form and reduced surgical time.⁹⁴ Forehead reconstruction is performed according to the surgeon's aesthetic judgment. Some centers advocate forehead overcorrection to accommodate growth and relapse.

Delaying anterior cranial vault remodeling and frontoorbital advancement as long as possible has the lowest risk of relapse, particularly in syndromic craniosynostosis patients.⁹⁵ Surgical correction of the posterior vault when patients are young has been advocated, because it provides adequate volume for the growing brain and delays frontal and midface surgery until the bone is more mature (Fig. 24-22).

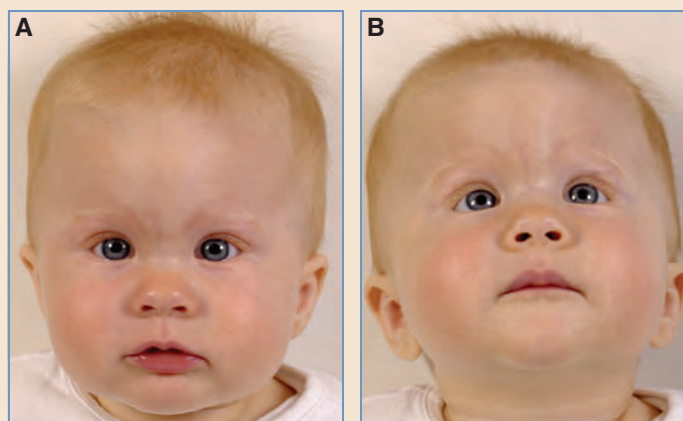


Fig. 24-22 A and B, This 7-month-old girl had metopic synostosis and underwent an anterior cranial vault reconstruction with bilateral frontoorbital advancement.

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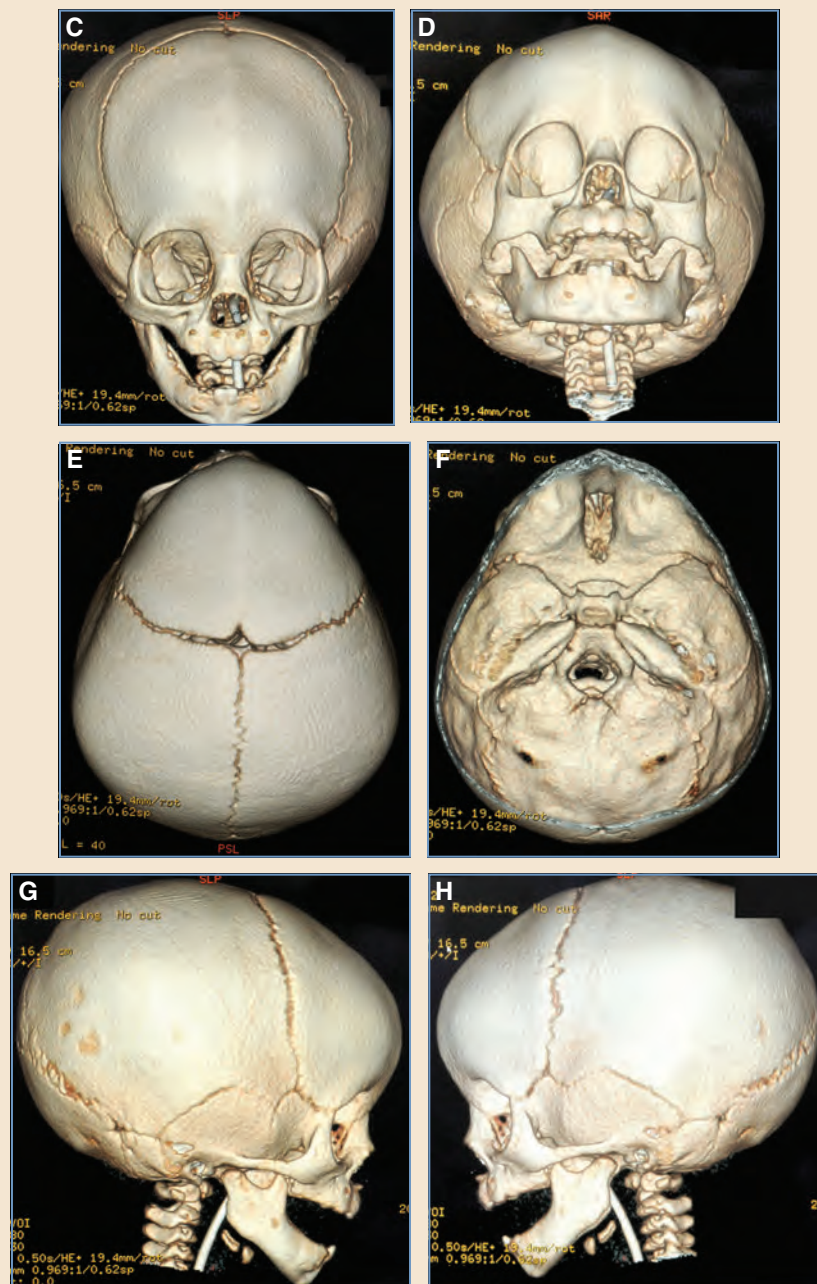


Fig. 24-22, cont'd C-H, Preoperative CT images.

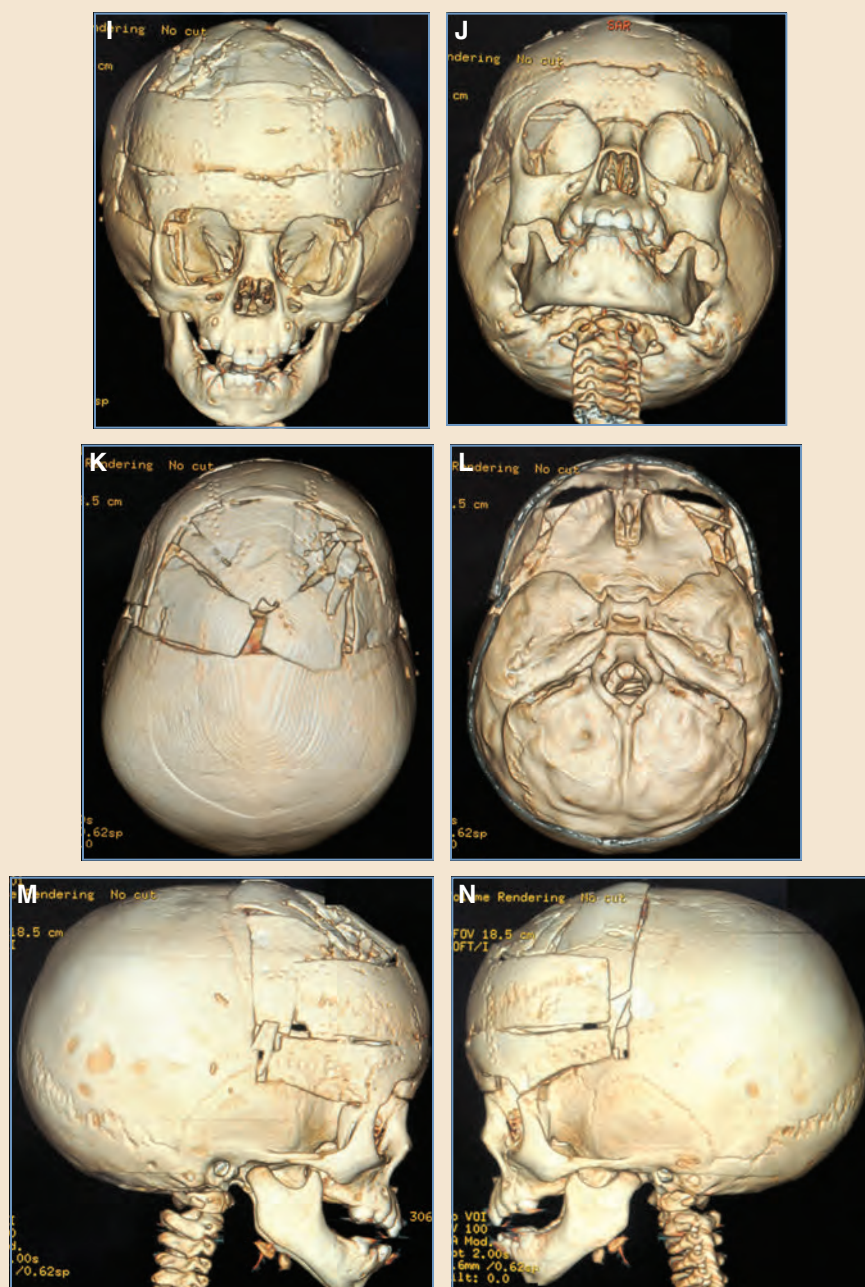


Fig. 24-22, cont'd I-N, Immediately postoperative CT images. The extent of surgery depends on the craniofacial deformity. This procedure involved removing the supraorbital rim and temporal attachment (bandeau) with subsequent reshaping. A symmetrical orbital aperture was created with a combination of contouring and reshaping maneuvers.

Continued

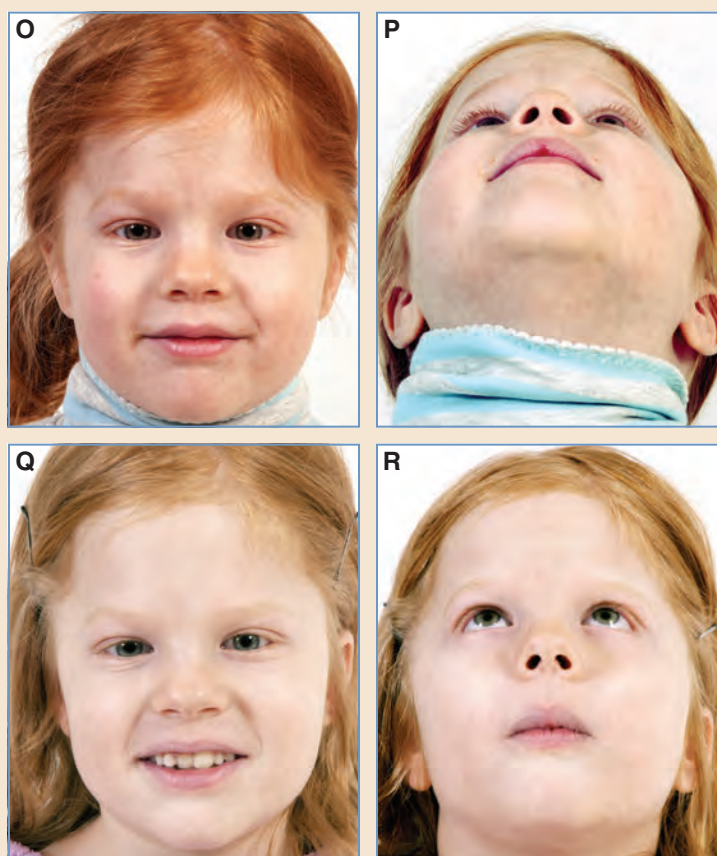


Fig. 24-22, cont'd O and P, Five years of age (4 years postoperatively). Q and R, At 7 years of age (6 years postoperatively), she has mild temporal recession.

Posterior Cranial Vault Reshaping

Indications

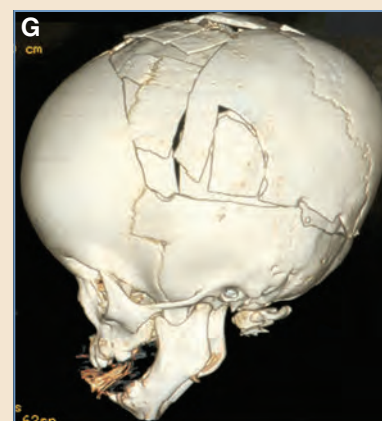
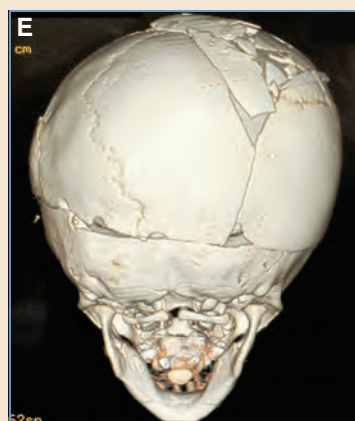
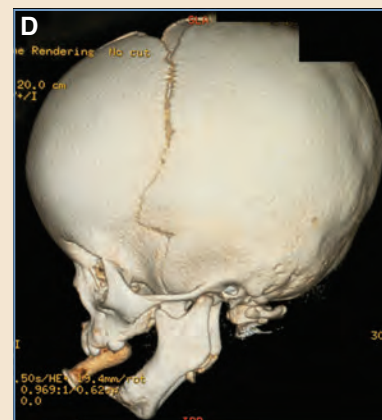
- Posterior plagiocephaly from lambdoid synostosis or a minor suture variant
- Turribrachycephaly from bicoronal synostosis
- Posterior fossa constriction

Procedure

Reshaping of the posterior cranial vault addresses deformities involving the posterior parietal and occipital regions that usually result from lambdoid synostosis or from the management of turribrachycephaly. Posterior cranial vault reshaping is not as commonly performed as anterior cranial vault reshaping and carries more risks because of the presence of the venous sinuses (the confluence of the sagittal and transverse sinuses, the torcula) and the mastoid air cells and because of the prone position of the patient, usually required for access. However, expansion and reshaping of the posterior cranial vault has become an area of first focus in the management of craniocerebral disproportion in syndromic craniosynostosis, in part because posterior expansion results in proportionately more intracranial volume (35%) compared with anterior frontal advancement (12% to 18%).⁹⁶ Furthermore, it may allow a delay in frontofacial surgery in syndromic cases, thereby helping to minimize relapse and the need for a repeat surgery. Generally,



Fig. 24-23 Posterior cranial vault reshaping is indicated for the rare cases of lambdoid synostosis that warrant surgical intervention, as part of a total cranial vault reshaping, and for expansion (in syndromic craniosynostosis). This patient with unilateral left lambdoid synostosis was treated with posterior cranial vault reshaping. **A-D**, Preoperative CT scans. **E-G**, Postoperative CT scans.



posterior cranial vault reshaping should be performed in patients 4 to 6 months of age or older, when bone is thick enough to withstand the application of fixation plates or distraction devices.

Techniques for posterior cranial expansion include a traditional craniotomy disassembly and reassembly cranioplasty (Fig. 24-23), posterior cranial vault distraction, and the use of springs.⁹⁷⁻¹⁰⁰ Cranioplasty techniques involve the use of bioresorbable plate fixation with reshaping and expansion of the cranial vault to maintain appropriate cranial form. This is an invasive technique and can be complicated by the presence of large, bony spicules on the endocranial surface that make bone removal hazardous.

Posterior cranial vault distraction has become a favored approach to posterior cranial vault expansion, because it is less invasive and allows titration of the degree of expansion and adjustment. It is a useful first-stage treatment for turriccephaly associated with syndromic craniosynostosis. The procedure involves a linear posterior craniectomy, followed by the application of small bone distractors. It does not incur the same risks of dural tears and sinus breach, because the bone is not removed (Fig. 24-24). The posterior cranial vault is slowly distracted at a rate of 1 mm per day to gradually expand the intracranial volume. This technique may also be used

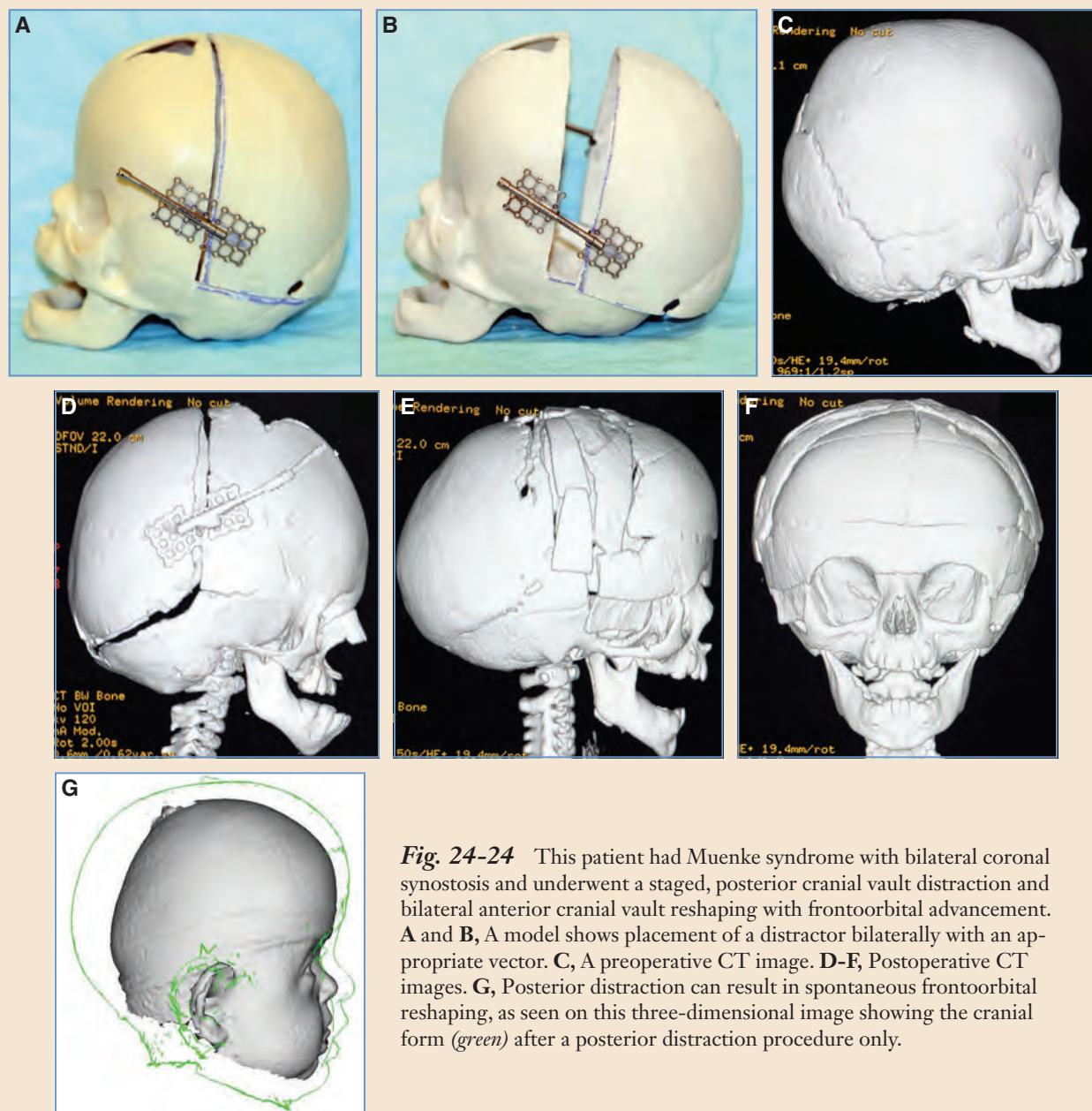


Fig. 24-24 This patient had Muenke syndrome with bilateral coronal synostosis and underwent a staged, posterior cranial vault distraction and bilateral anterior cranial vault reshaping with frontoorbital advancement. **A and B**, A model shows placement of a distractor bilaterally with an appropriate vector. **C**, A preoperative CT image. **D-F**, Postoperative CT images. **G**, Posterior distraction can result in spontaneous frontoorbital reshaping, as seen on this three-dimensional image showing the cranial form (green) after a posterior distraction procedure only.

to correct turricephaly by placement of the distractors in a downward vector. Disadvantages of distraction include the cost of the distractor devices, the inability to change the vector, and the need for a second procedure to remove the devices. Posterior cranial vault distraction has led to improvement in patients with Chiari malformation and may be a useful alternative to posterior fossa decompression in selected cases.¹⁰¹

Lauritzen and colleagues^{98,99} applied springs to the craniofacial skeleton as early as 1998. The spring was made of a metallic wire that was bent and placed across an osteotomy or open suture and anchored through holes in the calvarium or at the edges of the osteotomy. The spring separated the bony fragments until its inherent prebent shape was achieved or until it reached equilibrium with the surrounding tissue forces. The advantages of the spring were the relative ease of its application, the prevention of significant dissection and multiple osteotomies, and complete soft tissue coverage, which was not possible with distractors. The major disadvantages were the lack of control of the vectors and force applied, the need for a second surgery to remove the spring, and the possible associated complications, including dislodgement, skin penetration, and pressure sores.⁹⁹

Once intracranial volume is effectively expanded by posterior vault reshaping, the frontal region is corrected in a later stage, when skeletal maturity will decrease the risk of relapse (see Fig. 24-24).

Strip Craniectomy Techniques

Indications

- Scaphocephaly from sagittal synostosis in infants younger than 6 months of age
- Multiple-suture syndromic craniosynostosis

Procedure

Historically, removing a fused suture was thought to be the logical approach to managing the calvarial deformity of craniosynostosis in unlocking the growth potential of the skull. However, the standard method of a simple synostectomy of the involved suture was not always reliable for managing patients with scaphocephaly. More extensive craniectomy techniques developed in which additional bone was removed to allow normalization of the cranial vault. Dissatisfied with the results of these procedures, Jane and colleagues^{102,103} developed the pi procedure, which involved a strip craniectomy in the shape of the Greek letter π , followed by compression of the skull in an AP direction for side-to-side expansion.

Multiple variations on the pi procedure were developed, suggesting that this technique was not a panacea for all conditions. In 1991 Marsh et al¹⁰⁴ eloquently explained that with age, the ability of the bone to remodel under the influence of the growing brain diminishes. More extensive cranioplasty techniques were advocated in lieu of strip craniectomy techniques to adequately improve cranial dysmorphism from craniosynostosis.¹⁰⁴

This changed, however, with the advent of minimally invasive surgery techniques in the 1990s. In 1998 Jimenez and Barone¹⁰⁵ described the placement of an endoscope through small incisions to perform strip craniectomies safely in infants younger than 6 months of age. Rapid brain growth and thin bone allowed normalization of the cranial vault but required a powerful adjunct in the form of an external molding helmet to be worn for 4 to 12 months postoperatively. This technique has produced acceptable improvements in cranial dysmorphism and has been reproduced at multiple centers.¹⁰⁶

Currently, strip craniectomy techniques as an open or endoscopic-assisted approach are commonly performed. They have shown that improvement in craniofacial form is possible by

taking advantage of Moss' functional matrix theory.⁴ A variety of techniques are advocated (Fig. 24-25). Strip craniectomies are still performed in some units as a first-line treatment for managing elevated ICP in syndromic, multiple-suture synostosis cases in very young infants (younger than 3 months of age). This is temporizing at best because of rapid reossification, but it may buy time before the definitive surgery.

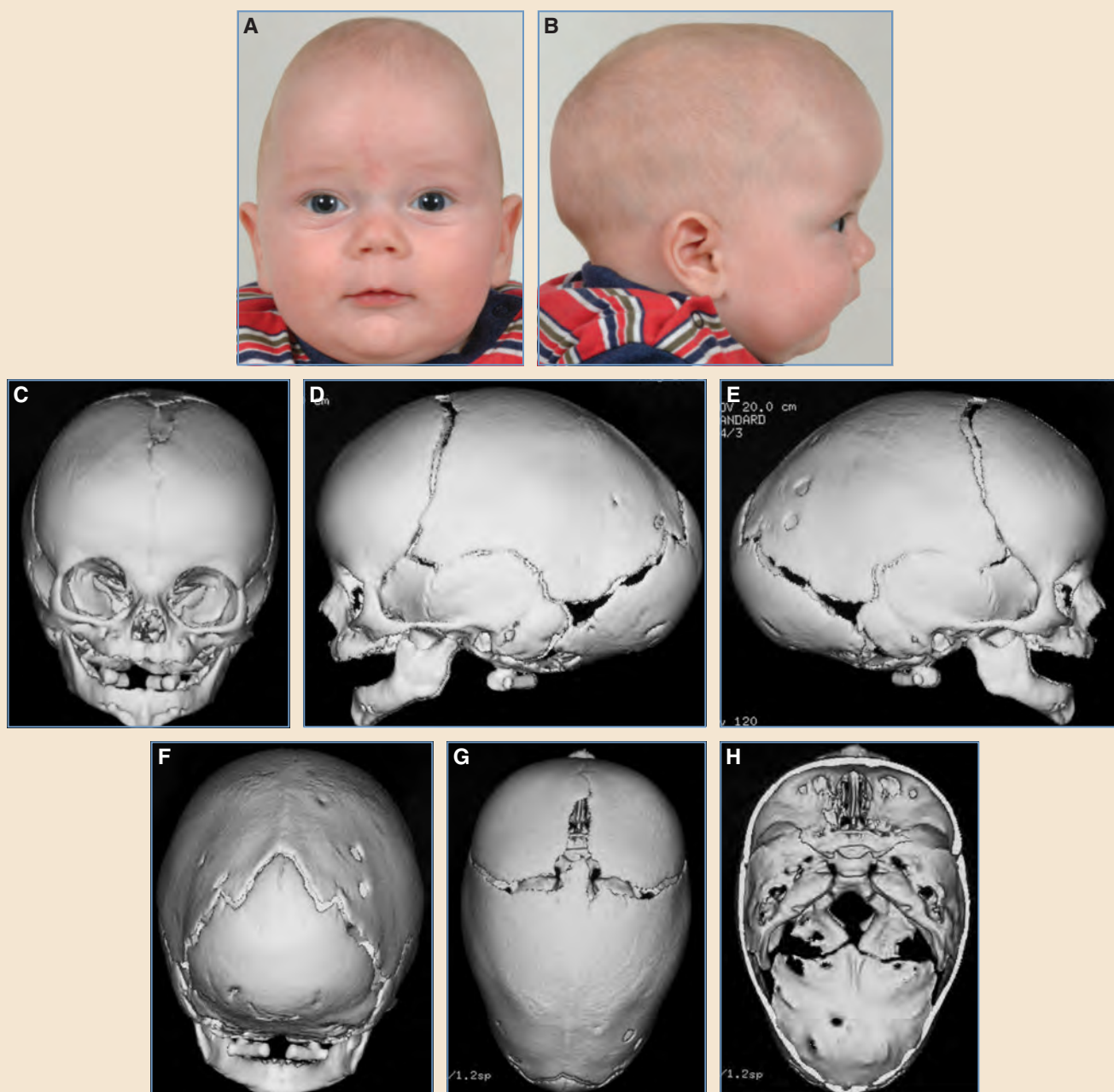


Fig. 24-25 A and B, This 5-month-old boy had scaphocephaly as a result of sagittal synostosis. He underwent an extended strip craniectomy. C-H, Preoperative CT images.

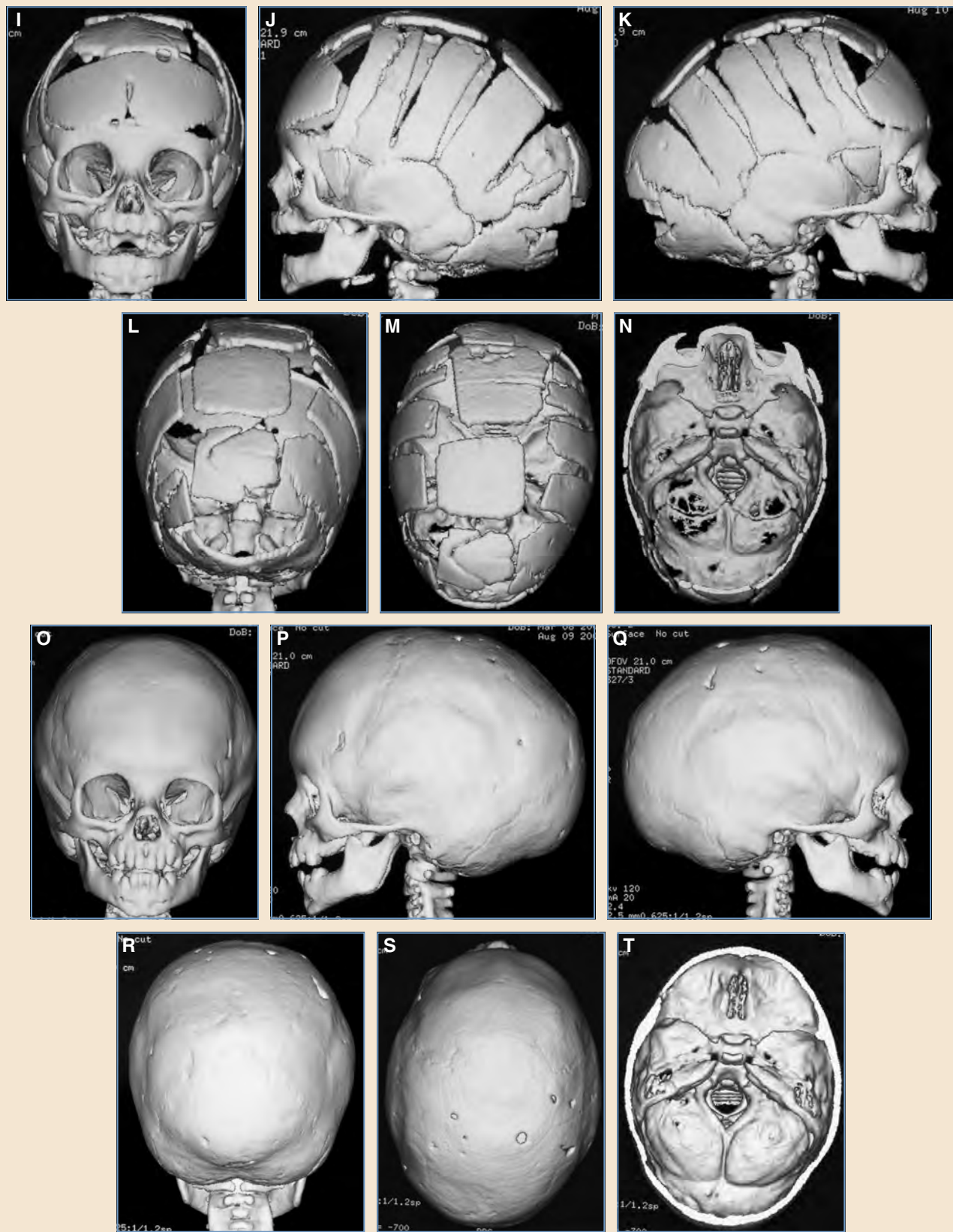


Fig. 24-25, cont'd I-N, Immediately postoperative CT images. O-T, One year postoperatively, his cranial shape has normalized.

Continued

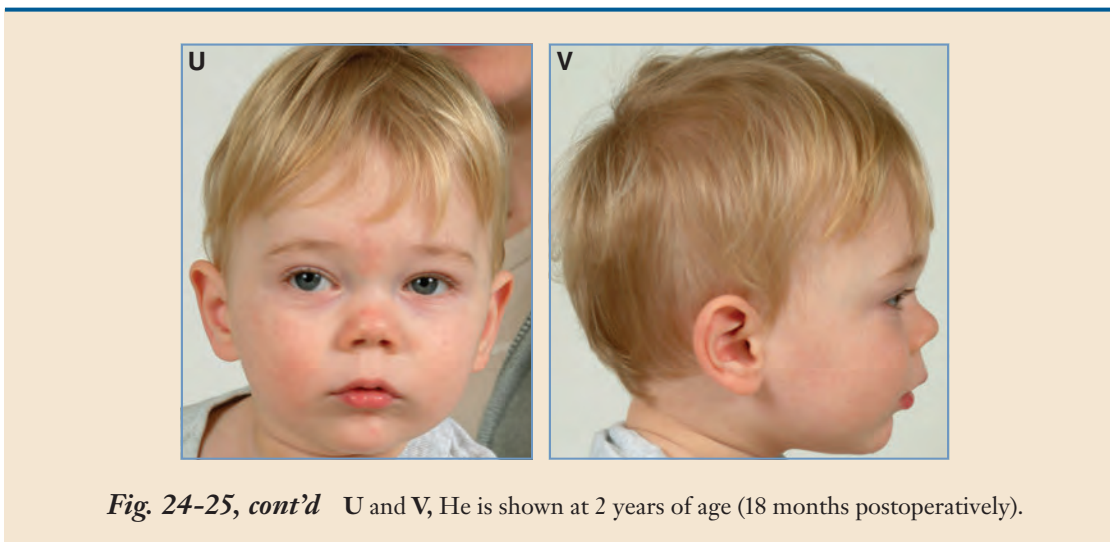


Fig. 24-25, cont'd U and V, He is shown at 2 years of age (18 months postoperatively).

Total Cranial Vault Reshaping

Indications

- Scaphocephaly from sagittal synostosis in infants 6 months of age and older
- Turribrachycephaly from bicoronal synostosis
- Multiple-suture synostosis

Procedure

Reshaping of the entire cranial vault is usually performed to correct cranial vault dysmorphism and craniocerebral disproportion associated with sagittal or multiple-suture syndromic cranio-synostosis. It is performed as a single or a staged procedure. Total cranial vault reshaping consists of a disassembly-reassembly cranioplasty technique using some form of fixation to maintain the cranial shape. The procedure is indicated for scaphocephaly correction in infants older than 6 months of age. Strip craniectomy techniques do not have the same degree of efficacy once the bone has thickened and brain growth starts to diminish. Ideally, if a frontoorbital bandeau is selected, it should be completed before the development of the frontal sinus to reduce the risk of contamination and infection.

For a total cranial vault reshaping, the patient is placed in a prone position for adequate access to the skull. However, this position makes airway management potentially more problematic and necessitates turning the patient intraoperatively (Fig. 24-26). The sphinx position allows access to the front and back of the cranial vault but increases the risk of air embolus and venous drainage problems, because the neck vessels are kinked with neck hyperextension.

Many of the techniques available for correcting skull deformities associated with sagittal synostosis are beyond the scope of this chapter. Surgeons should apply the concepts and principles of cranial expansion and reassembly to choose the best technique for each patient.

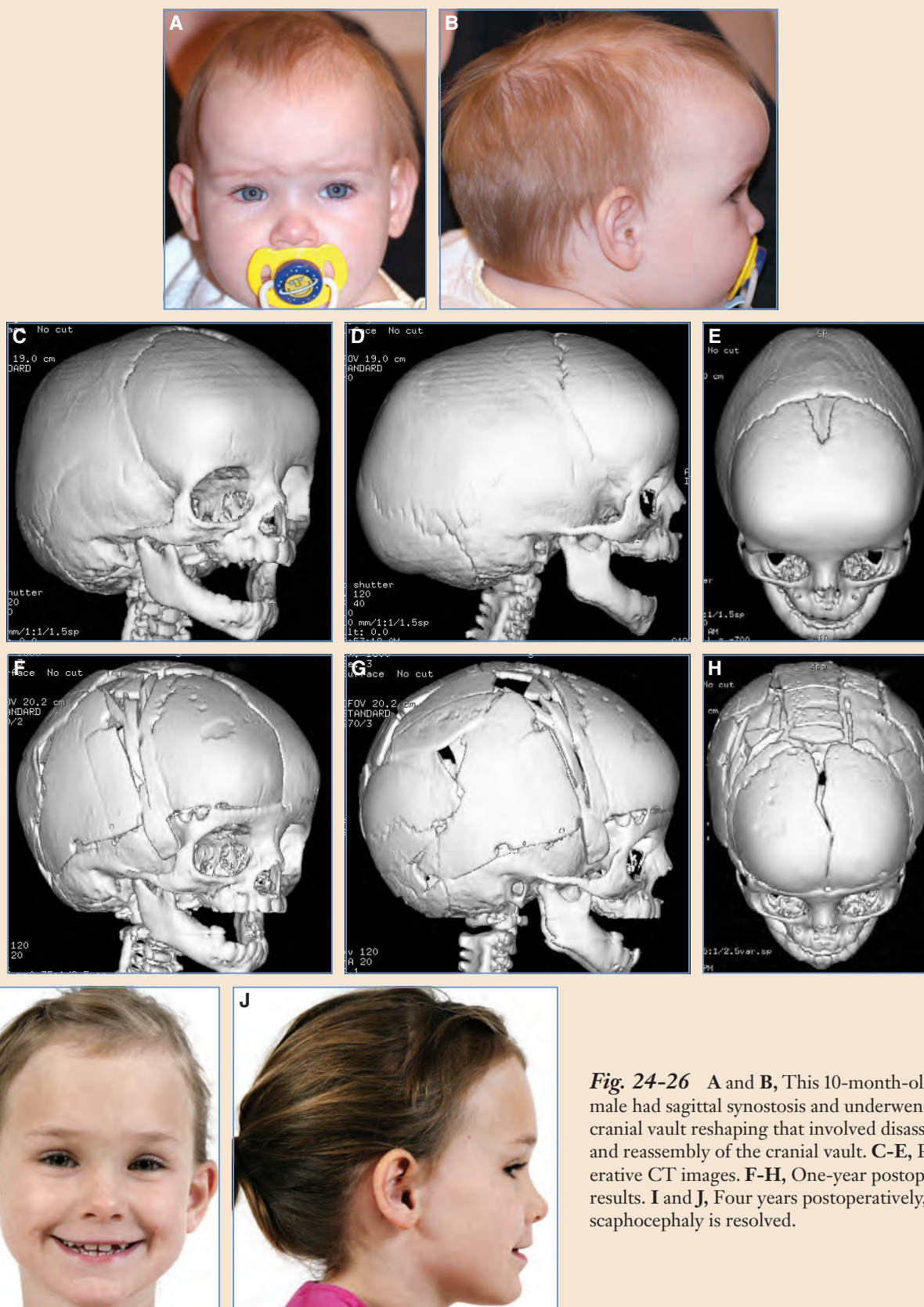


Fig. 24-26 A and B, This 10-month-old female had sagittal synostosis and underwent a total cranial vault reshaping that involved disassembly and reassembly of the cranial vault. C-E, Preoperative CT images. F-H, One-year postoperative results. I and J, Four years postoperatively, her scaphocephaly is resolved.

Other Techniques: Springs and Distractors

Springs have had wide application for managing many forms of single- and multiple-suture synostosis, in addition to posterior vault expansion surgery, discussed previously. Lauritzen et al⁹⁸ performed their original work at Sahlgrenska University Hospital in 1997. The procedure has been performed in centers around the world. In a young, pliable skull, a spring applied across a strip craniectomy will force the bone ends apart and remodel the adjacent cranial vault. Their use has evolved for managing patients with sagittal, metopic, and bicoronal synostosis. Critics of the technique cite the inability to control the degree and vector of expansion, possible erosion through the skin or through the dura, and difficulty removing the springs when they are enveloped by bone. However, once surgeons master the learning curve, the advantage of minimal intervention techniques and low-key technology, combined with experience, can produce satisfactory results.^{107,108}

Bone distractors have been applied to the cranial vault in patients with unicoronal, sagittal, and metopic synostosis. The advantages of not removing and reassembling bone fragments with fixation techniques are well-known. Disadvantages include the limited extent of correction, issues regarding vector control and cost, and the need for additional procedures to remove the devices. The optimal technique for the surgical management of craniosynostosis is not yet known, but having multiple options is invaluable.^{109,110}

Late Surgery

Craniosynostosis surgical intervention is recommended for patients 1 year of age or younger; however, many patients are not diagnosed until later in life. Several factors require consideration when treating older patients. The cranial bone is less malleable and more prone to fracture during manipulation. Cranial bone defect reossification is less robust and often requires split-skull bone graft during surgery or subsequent operations to fill the gaps.

Neurodevelopmental delays may not be identified until a child nears school age. Rarely, untreated craniosynostosis has caused intracranial hypertension and irreversible vision changes.

As mentioned previously, most single-suture craniosynostosis patients undergo a single intracranial procedure in the first year of life. However, because of facial asymmetry associated with unicoronal synostosis, bone defects, scar revisions, and areas of relapse, these patients are followed closely for several years and have the opportunity for additional surgery as needed (Fig. 24-27). The scope of management for syndromic patients is beyond the limits of this chapter but may involve a staged correction of hypertelorism, airway management with monobloc frontofacial advancement or LeFort III advancement, and definitive orthognathic surgery at skeletal maturity.

COMPLICATIONS

Surgery for craniosynostosis is considered a major, complex procedure performed on young infants with limited physiologic reserve. The potential for severe comorbidities (airway problems and intracranial hypertension) is high. These operations should be performed exclusively by a team of experienced health care professionals at a tertiary care center with adequate volumes. Despite the expertise and experience of such professionals, complications and, rarely, death occur from surgical intervention. Six of Tessier's fellows published the first report of complications in craniofacial surgery in 1979.¹¹¹ They described major morbidity and mortality rates of 16.5% and 1.6%, respectively.

Improvements in anesthesia and monitoring techniques, imaging, and technology resulted in shorter surgical times and significantly diminished mortality in large series. Most life-threatening

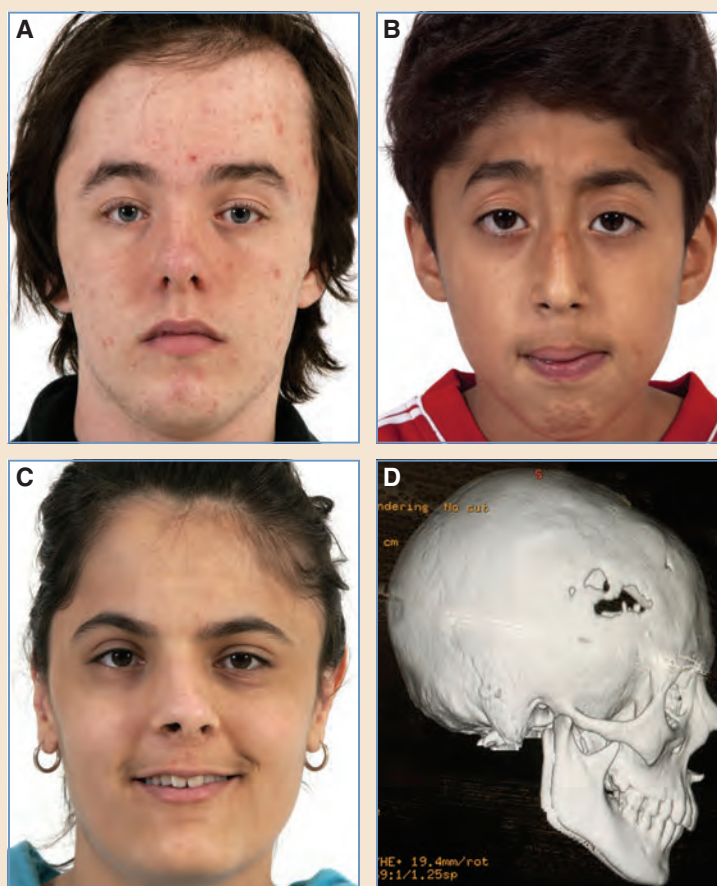


Fig. 24-27 These patients underwent a single procedure in infancy for treatment of a single-suture nonsyndromic synostosis. **A**, This patient presented with a mild right temporal hollow 10 years after correction of a right unicoronal synostosis. **B**, This patient had a bilateral superolateral orbital wall recession 11 years after correction of a metopic synostosis. **C**, This patient had residual facial asymmetry 10 years after a right unicoronal synostosis repair. She had discordance of orbital and dental midlines and periorbital asymmetry. **D**, Cranial defects after craniocorbital reshaping that persist in patients 5 years of age and older may require cranioplasty.

complications occur in the operating room and are related to blood loss and volume depletion, transgression of the venous sinuses, air embolism, and the effect of comorbidities such as cardiac disease and airway compromise. In a two-center review of 8100 intracranial and subcranial craniofacial procedures, the intracranial procedure major morbidity rate was less than 0.1%, and the combined mortality rate was 0.1%.¹¹² Subsequently, a Nationwide Inpatient Sample for 2004 through 2010 reviewed 17,788 hospitalizations for surgical repair in craniosynostosis patients up to 3 years of age.¹¹³ They reported a mortality rate of 0.8%. After hospitalization, 98% were discharged routinely. The five most frequently reported complications were hemorrhage (4.1%); iatrogenically induced complications, including accidental punctures, lacerations, and pneumothorax (3.1%); cardiac complications (0.7%); bacterial infections (0.7%); and respiratory complications (0.7%). No distinction was made between syndromic and nonsyndromic cases in either of these series.

In a recently published series of more than 700 intracranial procedures in nonsyndromic single-suture craniosynostosis patients, major complications occurred in 1.1% and minor complications in 3.5% of cases.¹¹⁴ Complications were increased with the presence of any comorbidity and in patients with metopic and sagittal synostosis.

A series of nearly 800 consecutive patients in Melbourne, Australia, had surgery for craniosynostosis from 1981 to 2010.¹¹⁵ Complications occurred in 14% of children, and 5.4% required a major revision surgery. Multivariate analysis helped to identify the following predictors of complications: multiple-suture and syndromic craniosynostosis, more recent surgeries, younger age (less than 9 months), spring-assisted cranioplasty, longer surgery, and greater transfusion volume. Patients with syndromic and multiple-suture craniosynostosis and those younger than 9 months of age at surgery had an increased risk of major revision surgery for regression to phenotype.¹¹⁵

Syndromic and multiple-suture synostosis patients are well-known to have higher rates of morbidity. McCarthy et al⁹⁵ documented their experience over 20 years at a dedicated craniofacial unit. They highlighted the complexity of care required with syndromic craniosynostosis patients and showed increased complication rates compared with those of isolated nonsyndromic single-suture craniosynostosis patients. Specifically, the incidence of major secondary procedures (36.8% versus 13.5%), perioperative complications (11.3% versus 5.0%), and secondary complications (44.7% versus 7.7%) were significantly increased in the syndromic cases.⁹⁵

Because corrective surgery requires significant periosteal stripping and craniotomies, major blood loss usually is expected. Strategies to minimize transfusion include hypotensive anesthesia, preoperative recombinant human erythropoietin, and intraoperative agents such as aminocaproic acid and tranexamic acid.¹¹⁶⁻¹¹⁹ Venous air embolus risk is well documented in children undergoing cranial procedures. Although hemodynamic compromise is rare, the incidence of asymptomatic air embolism may be as high as 82.6%, as assessed by precordial Doppler ultrasonography.¹²⁰ Lifting the cranial flap off the dura can result in a dural tear with CSF leakage. When this is recognized intraoperatively, direct repair with or without a pericranial patch is required. A fibrin sealant may be used as an adjunct for watertight closure. Infection is low at 2.5% to 3.2%.^{121,122}

The late complications are abnormal bone healing and impaired bone growth. Dura has osteogenic potential in patients up to approximately 18 months of age. Older children have an increased risk of persistent cranial defects compared with those younger than 1 year of age. Up to 5% to 20% of patients have incomplete ossification, correlating with age.¹²³⁻¹²⁵ Generally, for cranial defects larger than 2 cm at the initial repair, split calvarial bone grafts or particulate bone graft should be used. Subsequent defects may require a second-stage surgery with either additional bone graft or a cranioplasty. Multiple materials are available, including titanium mesh, polymethylmethacrylate, and more recently, custom computer-aided design and computer-aided manufacturing polyetheretherketone implants. Significant relapse of bone requiring additional remodeling after adequate internal fixation is uncommon. However, partial relapse has been observed. Causes can be multifactorial, including the age of the patient at the time of the surgery, the bone thickness or density, the potential molecular effects of the causative germ mutation, the severity of the deformity of the initial presentation, and the effect of cranial base restriction.

Unresolved Issues

The treatment of craniosynostosis has evolved significantly to become a safer and more predictable operation. Several issues are unsolved. Minimizing blood loss is paramount. Bony relapse and bone defects are not completely predictable. One of the biggest issues is the measurement of outcomes. No good measures are available to indicate the impact of major craniofacial surgery in children. This is an opportunity for the development of patient-reported outcome measures for the future.

FUTURE DIRECTIONS

The evolution of advances in craniofacial surgery over the past four decades has been profound. As medical care legislation increasingly focuses on outcomes-based assessments, the framework may shift toward increased efficiency and more predictable results. Computer-aided design and manufacturing may decrease surgical time and deliver improved precision and accuracy.¹²⁶ The next dimension in craniofacial surgery will focus on enhanced application of combined technologies such as imaging and robotic surgery. It will be an exciting time for those with more than a passing interest in this field.

KEY POINTS

- Craniosynostosis (premature fusion of the cranial sutures) may be classified by the number of affected sutures (single versus multiple), the cause (primary or secondary), nonsyndromic or syndromic, and the anatomic location (major sutures: sagittal, metopic, coronal, temporal squamosal, and lambdoid; minor sutures: frontosphenoidal and frontozygomatic).
- The metopic suture fuses by 8 to 9 months of age. All other sutures usually fuse in the third decade of life.
- Most single-suture craniosynostosis cases can be diagnosed based on a clinical assessment. Surgeons should understand that sutural growth allows bone to move at right angles to the suture (Virchow's law), and that single-suture craniosynostosis results in recognizable patterns of cranial dysmorphism. CT scans are not routinely required to diagnose major suture fusion.
- Sagittal suture synostosis is the most common form of single-suture craniosynostosis (1 in 2500 births), followed by metopic synostosis. Lambdoid synostosis is the rarest form of single-suture synostosis. Symmetrical single-suture synostosis (sagittal and metopic) is more common in males.
- Nonsyndromic craniosynostosis is much more common than syndromic craniosynostosis and accounts for up to 85% of cases. A multidisciplinary assessment includes craniofacial and plastic surgery, neurosurgery, ophthalmology, genetics, social work, nursing, and anesthesia assessments at a minimum.
- Suture fusion patterns in syndromic craniosynostosis cases often involve the coronal ring (paired coronal sutures, frontosphenoidal sutures, and sphenothmoidal sutures) and are frequently associated with midface retrusion, exorbitism, and hypertelorism.

Continued

KEY POINTS (continued)

- Craniosynostosis may be a component of up to 180 different syndromes, but Crouzon, Apert, Pfeiffer, Muenke, and Saethre-Chotzen are the more common syndromes seen. Syndromic craniosynostosis cases are quite complex and can be differentiated on the basis of an extremity examination. However, phenotypic differentiation is not always accurate and may require molecular genetic assessment (*FGFR*, *TWIST*, and *EFNB1*).
- Syndromic craniosynostosis cases require interdisciplinary team assessments, including CT, MRI, and vascular imaging to rule out hydrocephalus, Chiari malformations, and venous sinus anomalies.
- ICP may be increased in up to 50% of syndromic craniosynostosis cases and may result from craniocerebral disproportion, hydrocephalus, venous drainage anomalies, and obstructive sleep apnea. Detection of intracranial hypertension may be based on clinical findings (headache), although symptoms can be subtle or absent. Papilledema and bulging fontanelles may be present. ICP monitoring is considered to be the benchmark for assessing increased ICP.
- Surgical treatment of single-suture craniosynostosis may involve strip craniectomy techniques with molding helmet therapy, which is best performed in infants younger than 6 months of age. Patients older than 6 months of age have increased bone thickness and diminished brain growth, resulting in less cranial vault remodeling. Therefore surgical treatment in this age group requires the disassembly-reassembly cranioplasty techniques of anterior, posterior, or total cranial vault remodeling. A frontoorbital advancement is performed in patients with supraorbital rim retrusion or asymmetry.
- Surgical treatment of syndromic craniosynostosis is often staged and begins with cranial vault expansion and remodeling in infancy. Because of the higher reoperation rates, attention has recently been focused on addressing the posterior cranial vault initially and delaying treatment of frontofacial deformities. Delaying surgery may reduce the need for revision procedures. Frontofacial advancement, LeFort III, and orthognathic procedures are routinely carried out in syndromic craniosynostosis patients at specific developmental points depending on the indications.
- Syndromic and multiple-suture synostosis patients have higher rates of complications and morbidity.
- Despite best efforts, residual deformities can develop after surgical treatment. Therefore craniosynostosis patients should be followed at a recognized craniofacial center until the teenage years to provide the opportunity for correction of secondary deformities.

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25

Hypertelorism

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ypertelorism is a descriptive term commonly used to signify an increase in the distance between the bony orbits.¹ This abnormality is not a specific pathologic entity but is a trait shared by many conditions. Strictly defined, hypertelorism is an abnormally increased distance between *any* two paired body parts; for this reason Tessier et al^{2,3} preferred the term *orbital hypertelorism*, or *hypertelorbitism* (sometimes shortened to *telorbitism*). Despite this preference, the term hypertelorism has persisted as the descriptor for any increased bony interorbital distance. *Telecanthus* refers to an increased intercanthal (soft tissue) distance. Although telecanthus always accompanies hypertelorism, the reverse is not always true—in some instances, there may be an increased intercanthal distance without true hypertelorism; for example, blepharophimosis or laterally displaced nasoethmoid fractures with disruption of the medial canthal tendon.

CONDITIONS THAT CAUSE HYPERTELORISM

In embryologic development, the optic vesicles arise laterally by the fourth gestational week; their medial migration occurs both as a primary process and secondarily as a result of rapid expansion and broadening of the cerebral hemispheres and temporal lobes. The final position of the optic vesicles is set at about the ninth week of gestation. Hypertelorism results when any condition interrupts this medial movement or when some midline structure, such as the ethmoid sinus or the brain, expands abnormally and pushes the orbits apart. The most common causes for hypertelorism are listed in Box 25-1; however, these conditions are not exclusive and may overlap. Given that hypertelorism is a phenotypic observation common to multiple conditions and syndromes, it is not surprising that there has not been a single gene found responsible for this trait. Instead, multiple gene mutations have been correlated with hypertelorism.

Box 25-1 Conditions Associated With Hypertelorism

- Craniofacial dysostosis
- Encephaloceles
- Facial clefting
- Frontonasal and craniofrontonasal dysplasia
- Miscellaneous: tumors; Greig, Opitz, and other syndromes
- Trauma
- Amnion rupture sequence



Fig. 25-1 CFND includes coronal synostosis (left-sided in this patient) in association with hypertelorism.

A relatively common cause of significant hypertelorism is *craniofrontonasal dysplasia sequence* (CFND). This is an atypical X-linked syndrome with variable penetrance; it occurs more often and is usually more phenotypically severe in females than in males. In most cases, this condition has been found in association with a mutation in the ephrin B1 (*EFNB1*) gene.⁴ Affected children present with a combination of both coronal synostosis (unilateral or bilateral) and frontonasal dysplasia, resulting in an asymmetrical hypertelorism that is frequently accompanied by a widow's peak and broad nasal tip.⁵⁻¹⁰ (Fig. 25-1). *Frontonasal dysplasia* is a condition similar to CFND except with no associated craniosynostosis; multiple subtypes have been identified, many of which are in association with mutations in the *ALX* homeobox genes 3, 4, and 1.¹¹ Hypertelorism may also result from facial clefting, amnion rupture sequence, or in association with encephaloceles. In particular, midline facial clefting may be associated with other midline intracranial anomalies. There may also be some correlation between midline facial clefts (Tessier number 14) and encephaloceles.¹² Encephaloceles may be the one exception to the belief that in most cases of hypertelorism, the distance between the optic canals is either normal or only slightly increased.¹³ When present, encephaloceles may exit the cranial cavity through one of multiple locations. When the encephalocele exits through the foramen cecum (a frontal encephalocele) or through the floor of the skull (a basal encephalocele), midline migration of the orbits is usually affected, resulting in *clinical hypertelorism*.

Significant facial trauma most often results in telecanthus, but it may also cause hypertelorism; however, in these cases, a component of vertical globe dystopia and significant asymmetry often accompanies the increased interorbital distance. Patients with craniofacial dysostoses (for example, Apert, Crouzon, and Pfeiffer syndromes and others) may present with a mildly increased interorbital distance (probably from compensatory lateral temporal expansion resulting from the inhibition of frontal growth). Finally, many miscellaneous causes, such as fibrous dysplasia, tumors, and syndromes such as Greig, Opitz, and others, share hypertelorism as a common phenotypic component.

PERTINENT ANATOMY

Various methods of measuring the interorbital distance have been reported that are based on absolute distances and ratios of different anatomic structures: intercanthal distance, interpupillary distance, interdacryon distance, canthal index, circumference-interorbital index, intercanthal palpebral fissure index, and other parameters.^{14,15} The interdacryon distance is probably the most commonly used measurement. Normal values for interorbital distance have been described based on anatomic and radiologic measurements¹⁶⁻¹⁹ (Table 25-1).

Günther²⁰ described the following grading system for hypertelorism in adults: first-degree (30 to 34 mm), second-degree (35 to 39 mm), and third-degree (40 mm or more), and it has been suggested that the surgical approach should vary based on severity. The obvious drawback to this classification scheme is that it is only appropriate for fully grown individuals. For a growing child, it is more practical to base treatment decisions on a percentage increase in interorbital distance instead of using absolute numbers; for example, grade I (up to 125% of normal/average); grade II (between 125% and 150%); and grade III (greater than 150%). A classification system for hypertelorism has also been proposed based on the location of the maximum width of the ethmoid sinuses, as viewed in the axial plane (anterior, medial, or posterior). Cases involving posterior ethmoidal widening are the most difficult to correct; surgeons must be cautious when reducing the posterior ethmoid sinuses, which lie close to the entrance of the optic canal.²¹ Despite the possible surgical implications, this classification system has not been widely adopted. Nevertheless, knowledge of the anatomy of the presenting deformity is critical to achieving a safe, satisfactory correction.

Table 25-1 Average Interorbital Distances and Intercanthal Distances According to Age

Age (yr)	IOD (mm)	ICD (mm)
Birth	16	20
1	18.5	25
3	21	26
5	22	27
7	23	28
12	26	29
Adult	27	30

ICD, Intercanthal distance; IOD, interorbital distance.

PREOPERATIVE ASSESSMENT

Hypertelorism does not always require surgical treatment. In fact, many consider mild hypertelorism an attractive trait. An examiner's perception of interorbital distance may be affected by the height of the nasal dorsum: individuals with a hypoplastic nasal dorsum may appear to have an increased interorbital distance, even if the measured distance is in the normal range. In addition, epicanthal folds, exotropia, widely spaced eyebrows, narrow palpebral fissures, and dystopia canthorum may contribute to an illusion of hypertelorism.¹³ It has been the senior author's experience (J.A.F.) that correction of hypertelorism is almost never required with craniofacial dysostoses. This is because the associated nasal and midfacial hypoplasia significantly contributes to the appearance of hypertelorism, but when this is appropriately corrected, the improvement in the patient's appearance is sufficient to avoid the need for an intracranial hypertelorism correction.

Axial, coronal, and three-dimensional CT scan reconstructions are important tools for surgical planning. Many surgeons find three-dimensional models constructed from CT data helpful to determine osteotomy locations and plan the movements of bony fragments.²²⁻²⁴ For cases involving encephaloceles, MRI scans provide important data for the neurosurgical aspects of the repair and help to determine the boundary between the disorganized encephalocele and the organized functional brain. Finally, preoperative ophthalmologic evaluations provide baseline visual data to which subsequent evaluations may be compared.

SURGICAL TECHNIQUE

Timing

The timing of surgical intervention has engendered some degree of controversy among craniofacial surgeons and ideally should be based on several factors. Children who present with encephaloceles or facial clefts involving absent skull will typically require earlier treatment. Extreme care must be taken in repairing telorbitism arising from basal encephaloceles, because these may contain the pituitary gland and/or the hypothalamus, and resection of the encephalocele may lead to panhypopituitarism. Children with severe hypertelorism may be unable to develop stereoscopic vision if treatment is delayed (although the efficacy of treatment with respect to the development of stereoscopic vision has not been well studied). One issue regarding surgical timing relates to what effect growth will have on the final result. Some reports have suggested that growth is not affected by surgical treatment, which opens the door to earlier surgical intervention, whereas others suggest that surgery has a negative impact on growth.^{9,15,19,25-29} It seems intuitive that the larger the surgical procedure and the younger the patient, the more likely that growth centers will be affected and that subsequent growth will be diminished. Most experienced surgeons will delay surgical correction until a significant amount of growth has occurred, yet before any serious psychological issues have developed (which usually occurs at 6 to 8 years of age, depending on the individual), which seems to result in better outcomes. It has also been suggested that surgical interventions before 8 years of age may lead to higher rates of bony interorbital relapse.²⁷

The treatment of children affected with CFND, one of the more common causes of severe hypertelorism requiring surgical intervention, presents a more complicated timing consideration for surgery. The combination of hypertelorism (ideal correction delayed until 8 years of age) with unicoronal or bicoronal synostosis (ideally corrected before 18 months of age) makes a staged-surgery strategy necessary. Kawamoto et al⁹ described an operative algorithm for CFND based on their 30-year experience; they advised earlier treatment of the craniosynostosis at 6 months (although today, we would argue for a later correction: between 11 and 18 months of age), followed by a hypertelorism correction after the central incisors erupt at 6 to 8 years of age. In their series, the only patients requiring secondary hypertelorism corrections were those operated on earlier than 5 years of age.

Choice of Procedure

Tessier³⁰ opined that surgical correction for telorbitism is essentially surgery of the nose. Although correction of the associated nasal deformity is a critical finishing step in the treatment of hypertelorism, medial transposition of the globes is nevertheless a significant surgical undertaking. Numerous osteotomies have been described for correcting hypertelorism. To decide which of these techniques is the best approach, it is helpful to consider the underlying cause of the hypertelorism, the degree of separation, and the patient's age. The surgical treatment of hypertelorism is both one of the more complex and least commonly performed operations by craniofacial surgeons. The following description of the surgical techniques is presented as a general overview and should not be considered a surgical atlas.

Two-Wall, Three-Wall, and Four-Wall Osteotomies

Treatment of the mildest degrees of hypertelorism may be accomplished by performing *two-wall osteotomies*—superior and inferior osteotomies of both the medial and lateral orbital walls. The medial orbital walls are infractured or medially translocated after removal of a central strut of nasal dorsum, perpendicular plate, and posterior ethmoid sinuses, sufficient to permit medial globe translocation. The lateral orbital walls are then shifted medially, which moves the globes (Fig. 25-2). The medial movement of the lateral walls is responsible for reducing the distance between the globes in each of the different types of hypertelorism corrections. Simply shifting the medial walls more medially without moving the lateral walls will have no significant effect on globe position. The two-wall technique may be performed completely extracranially, avoiding the need for neurosurgical assistance, but care must be taken to preserve symmetrical orbital widths after translocation.

The *three-wall osteotomy** technique preserves the relationship of the orbital roof (or the orbital floor) to the medial and lateral walls, which in turn maintains the orbital width during translocation. If the width of the piriform aperture is normal, then any medial movement of the orbits will compress the nasal passages; therefore bone removal from the edges of the piriform aperture may be necessary to preserve the nasal airway. One advantage of the *U-shaped osteotomy*,

*The terms *three-wall* and *four-wall osteotomies*, although useful descriptors, are misnomers, because the orbit is composed of only two walls, along with a roof and a floor.

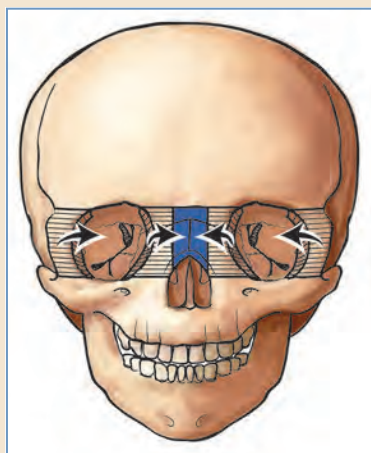


Fig. 25-2 Smaller orbital translocations can be achieved with two-wall osteotomies. The medial orbital walls are infractured, and the lateral orbital walls are moved medially, taking care to maintain normal orbital width.

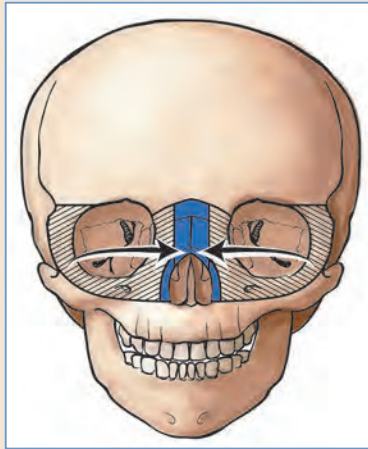


Fig. 25-3 U-shaped osteotomies include the orbital floor, with the medial and lateral walls as a single unit. These osteotomies exclude the orbital roof, avoiding the need for an intracranial approach.

Fig. 25-4 Reverse U-shaped osteotomy includes the orbital roof with the medial and lateral walls, eliminating the orbital floor osteotomy. This procedure requires a craniotomy, but it may be technically easier than the U-shaped osteotomy.

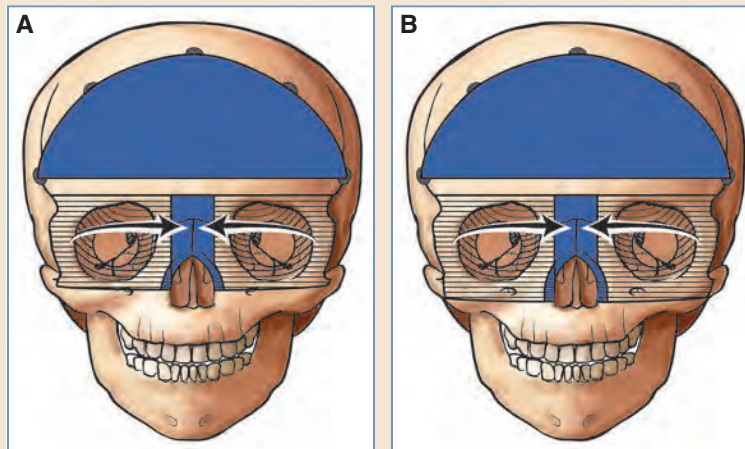
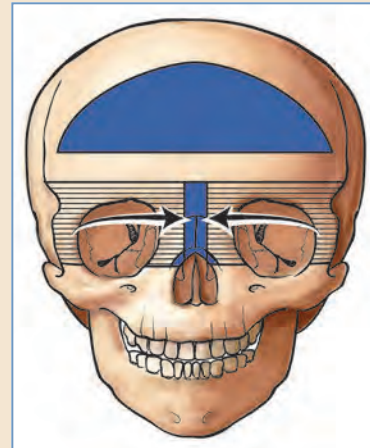


Fig. 25-5 The traditional four-wall osteotomy can be made either **A**, above the infraorbital foramen (in older children), or **B**, below the infraorbital foramen. However, osteotomies going below the infraorbital fissure may increase the risk of dental root injury.

which preserves the orbital floor (Fig. 25-3), is that this approach is completely extracranial. However, a disadvantage of this technique is that the infraorbital osteotomy can be difficult to perform without injury to the infraorbital nerve, lacrimal fossa, and unerupted tooth buds, especially in younger children. The *reverse U-shaped osteotomy* has several advantages: it leaves the orbital roof intact and is technically less challenging, the osteotomies are more easily visualized, and the maxilla is left intact (Fig. 25-4). The primary disadvantage of this supraorbital approach is the need for intracranial access.

Four-wall osteotomies (“box osteotomies”) are generally reserved for the most severe cases of hypertelorism and are generally undertaken in skeletally mature patients (Fig. 25-5). With this technique, osteotomies are made above the orbital roof and below the orbital floor, and the orbit is circumferentially cut in the coronal plane posterior to the midportion of the globe. Medial translocation is achieved by removing a central section of nasal bone, perpendicular plate, and posterior ethmoid sinuses. This approach provides complete mobilization of the orbits, allows more significant translocations, and may result in greater final stability.

Facial Bipartitions

Facial bipartitions are probably the most frequently performed hypertelorism correction technique for children, especially those who have any condition that has produced a midline separation of the face (for example, facial clefts and encephaloceles). One reason for this is that this technique avoids the need to perform a horizontal maxillary osteotomy, which might damage the roots of the teeth during the mixed dentition phase. This technique requires intracranial exposure, and in addition to bringing the orbits closer together, lowers the midline of the maxilla through a rotational movement of the lateral facial halves (Fig. 25-6).

A facial bipartition is also useful for children with significant midline vertical shortening of the maxilla, palate, and upper dental arch.^{31,32} Osteotomies are performed through the orbital roofs, medial and lateral walls, and floors in the coronal plane, as with a four-wall osteotomy. However, unlike the four-wall osteotomy, no horizontal maxillary cuts are made below the orbit. Instead, a pterygomaxillary disjunction is performed sagittally to allow the hemimaxilla to

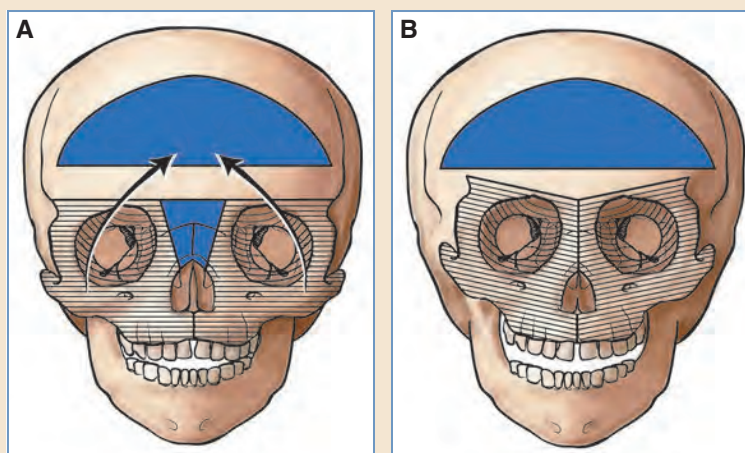


Fig. 25-6 Facial bipartition results in both **A**, a medial translocation of the orbits and **B**, flattening of the upper occlusal plane by rotating the facial halves through a lower central maxillary axis.

move in continuity with the orbit. This disjunction may interfere with future facial growth (and subsequent eruption of molars), depending on the child's age. Although modifications have been described that are intended to avert growth inhibition, the subsequent postoperative preservation of normal facial growth remains undocumented.³³ The facial bipartition technique creates larger bone segments for translocation than does the four-wall osteotomy technique, which may enhance final stability. In extremely rare instances of severe hypertelorism with significant increases in interoptic nerve distance, a staged correction should be considered to avoid the potential loss of vision from optic nerve tension after significant movement of the orbits (Figs. 25-7 through 25-9).

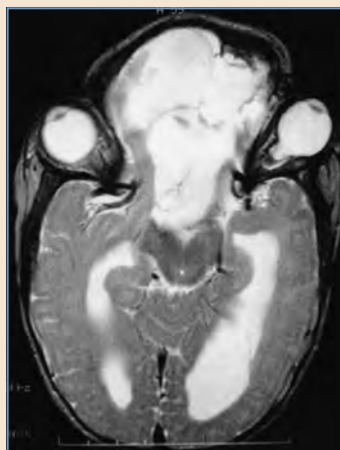


Fig. 25-7 Large encephaloceles may create a significant diastasis between the optic nerves, which is not typically seen in most other causes of hypertelorism.

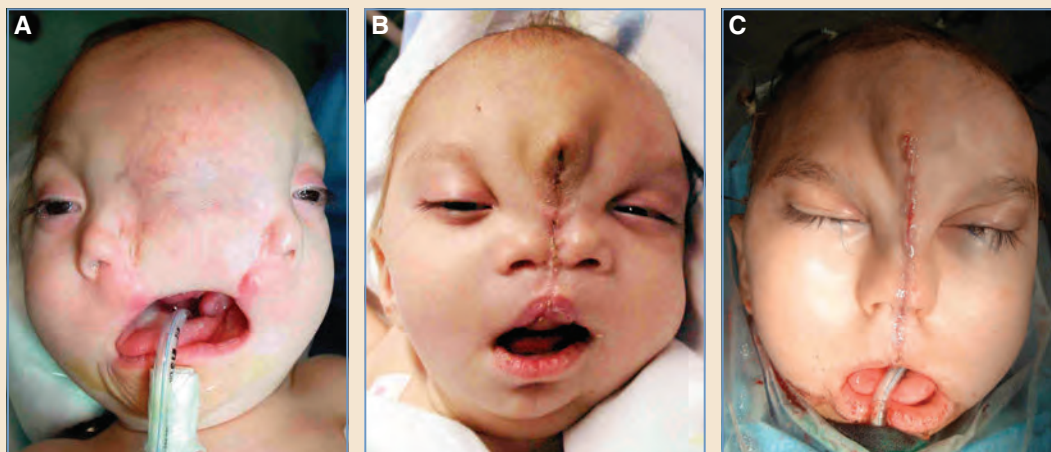


Fig. 25-8 **A**, A child with hypertelorism (resulting from an encephalocele) is seen preoperatively. A staged correction was planned to reduce the substantially increased distance between the optic nerves, based on concern for loss of vision. **B**, The same child is shown after the first stage of a bipartition procedure. A superior dog-ear is left unresected to limit the length of the skin incision. **C**, The child is shown after the second stage of the bipartition procedure.

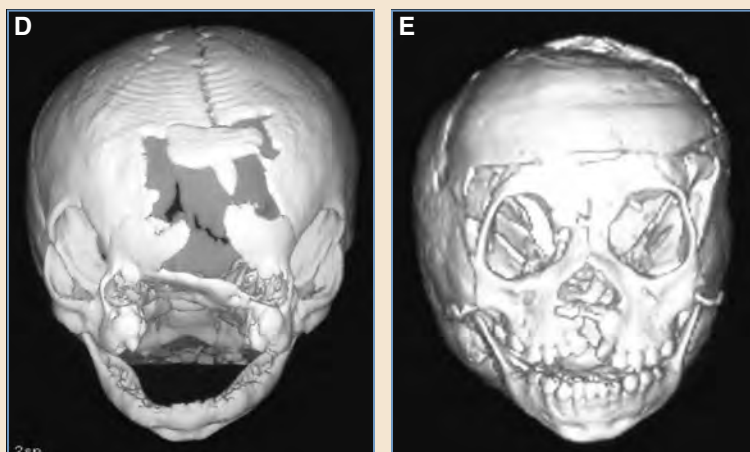


Fig. 25-8, cont'd D and E, Preoperative and postoperative three-dimensional CT scans.

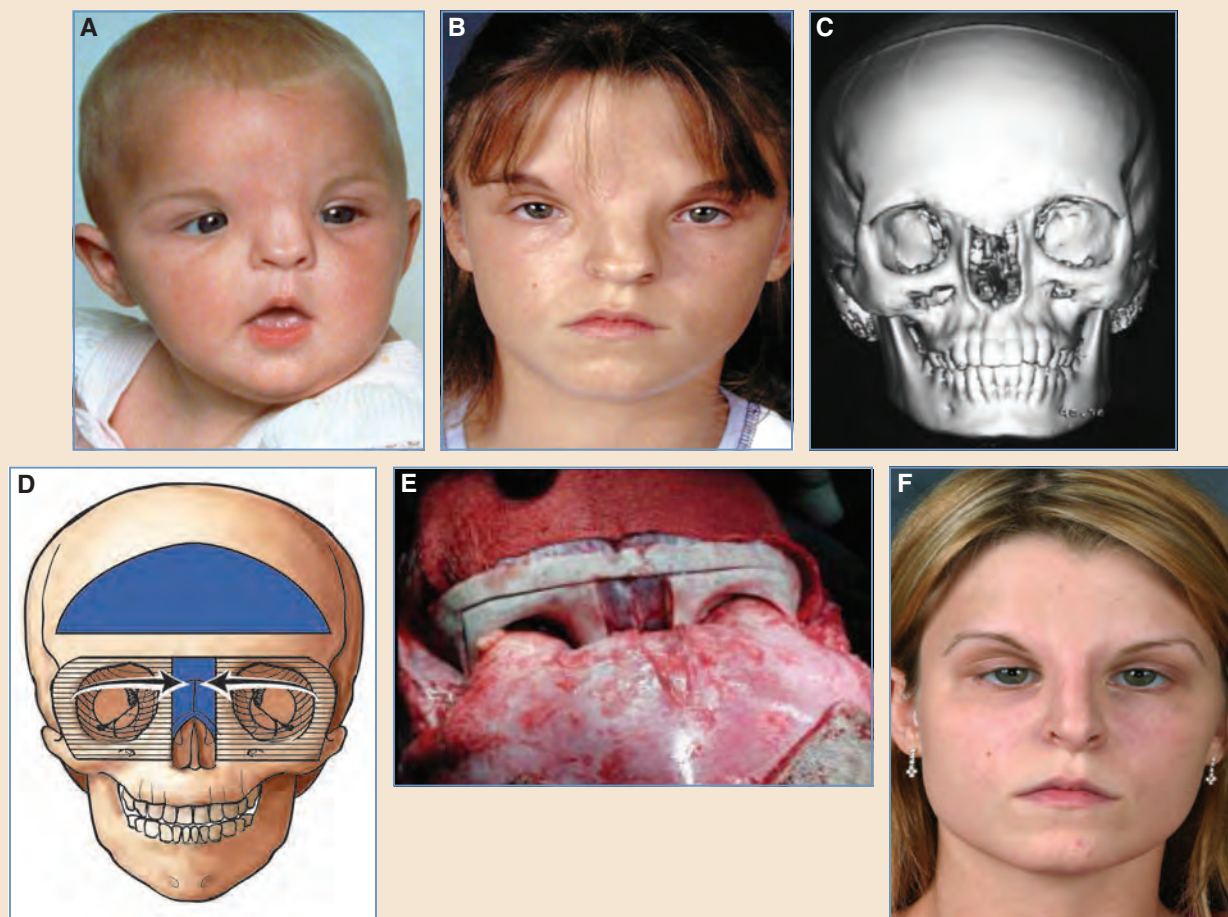


Fig. 25-9 Typical hypertelorism (see Chapter 35 for a case of atypical hypertelorism). **A**, Patient at infancy. **B**, Patient at 14 years of age with asymmetrical hypertelorism (atypical craniofacial cleft, bilateral Tessier 12-13). **C**, Preoperative three-dimensional CT scan. **D** and **E**, She underwent “box” orbital osteotomies at 15 years of age, followed by a nasal dorsal rib graft at age 16. **F**, Postoperative image at 17 years of age. The excess forehead skin was managed with an internal cinch suture. Further surgery is planned to aesthetically define the medial supraorbital rim and brow. (Courtesy of Pravin Patel, MD.)

TECHNICAL ASPECTS

We recommend a conservative approach to treat milder hypertelorism, not only because many individuals consider mild hypertelorism an attractive trait, but also because of the unfavorable risk-benefit ratio. Significant risks are associated with these major procedures, and in milder cases only limited enhancements in appearance can be expected. This philosophy restricts treatment to more severely affected individuals, and therefore the choice of osteotomies is narrowed to either a bipartition or a four-walled technique. Surgeons without significant pediatric congenital craniofacial experience are well advised to seek the assistance of more experienced surgeons when faced with a child who has significant hypertelorism.

Moderate-to-severe hypertelorism corrections begin with a low frontal craniotomy following an extensive dissection of the nasal bones, orbits, anterior zygoma, and maxilla. The low frontal craniotomy should be sufficiently wide to provide a clear view of the entire orbital roof and anterior tip of the temporal lobes, which need to be retracted while lateral orbital osteotomies are performed. An upper buccal sulcus approach is necessary with facial bipartitions to separate the midline of the palate or when performing box osteotomies if the low horizontal maxillary osteotomy is planned below the infraorbital foramen. After the frontal craniotomy, the width of the midline nasal resection is determined and marked, and cuts are made with a short saw blade to excise the central vertical strip of nasal bones; removing the posterior perpendicular plate of the ethmoids may help to further reduce the intraorbital distance. Osteotomies are made across the orbital roof anteriorly and then lateral to the lateral orbital walls to include as much of the posterior lateral wall as possible. Next, a horizontal cut is made below the inferior orbital rim, retaining sufficient bone to provide inferior stability, yet avoiding injury to the inferior orbital nerve. This low cut must continue medially into the vertical midline gap, which was created by removal of the central portion of the nose. Osteotomies are completed along the orbital floor anteriorly and down the medial orbital wall (or the medial wall can be removed).

For a bipartition, the low horizontal cut across the maxilla is replaced by continuing the lateral orbital osteotomy, which preserves as much of the lateral orbital wall as possible, into the pterygoid fossa to separate the pterygomaxillary junction. If the maxillary midline is intact, a limited upper buccal incision allows access to the hard palate. The hard palate is split along the midline with a narrow osteotome after the nasal mucosa is freed up along the floor of the nose and the inferior portion of the septum is detached from the vomerine groove. The two orbital boxes are then medially translocated.

With the bipartition procedure, each lateral facial segment is translocated, with the axis of rotation centered above the central incisors. We have found it helpful to place a temporary wire around the central incisors to prevent the creation of an unsightly gap between these teeth. In our practice, the medial orbits are held together with a monofilament absorbable suture such as 2-0 PDS, and the frontal bone is subsequently reattached with the same suture material. Our preference for absorbable suture material rather than resorbable sutures or metal plates is based on ease, speed of fixation, and low infection rates.³⁴

Medial canthopexies are then performed, also with the use of PDS sutures. The decision to primarily bone graft the nasal dorsum depends on the dorsal height of the paired nasal bones. After the coronal incision is closed, consideration is given to resecting the nasal skin through a midline excision. If skin needs to be removed, acceptance of a small dog-ear following a limited excision length is recommended, because this will flatten out over time.

POSTOPERATIVE CARE

Children undergoing hypertelorism correction require highly skilled postoperative pediatric intensive care monitoring. We do not use surgical drains or postoperative bandages, and children are routinely extubated in the operating room. Frequent neurologic checks and visual evaluations are performed, in addition to careful monitoring of hemoglobin levels, acid-base status, and serum electrolytes (observing for hyponatremia, indicating the possibility of a syndrome of inappropriate antidiuretic hormone, and hypokalemia, which may be induced if sodium bicarbonate is administered to correct acidosis). Typically, children require one to two nights of intensive care observation before they are sufficiently stable to permit safe transfer to a general unit. When hypertelorism corrections are performed in conjunction with basal encephalocele repairs, postoperative hormonal monitoring of pituitary and hypopituitary function is necessary.

COMPLICATIONS

Although not typically considered a complication per se, the inability to achieve a reasonably normal long-term appearance after surgical correction of hypertelorism is probably the most commonly encountered adverse outcome. In most instances the primary motivating factor to undertake a hypertelorism correction is to improve appearance, yet the ability to actually achieve a normal postoperative appearance can be extremely challenging. The reasons for suboptimal outcomes remain a matter of ongoing debate.^{9,15,26-28} Despite the commonly seen suboptimal hypertelorism correction results, most patients and their families report satisfaction with their surgery.^{28,35}

Few centers have the facilities and experience to provide these types of treatment, and even fewer centers have the volume of patients necessary to acquire the requisite experience to reproducibly and safely correct these rare anomalies. Given the rarity of this condition, there have been only a few published series of hypertelorism corrections coming from the busier centers; for this reason, complication rates are difficult to ascertain.

Among the objectively traceable complications, infection probably ranks as the most frequently encountered adverse outcome. Intuitively, infection rates for hypertelorism corrections should be higher than rates after isolated craniosynostosis corrections, because hypertelorism-associated osteotomies routinely traverse sinus cavities. Most craniofacial infections do not present clinically until about 10 days after surgery, but if prompt treatment is undertaken, bone loss may be minimized.³⁶ Brain injury is extremely uncommon, aside from the treatment of hypertelorism resulting from basal encephaloceles. In addition to infections, cerebrospinal fluid leaks, anosmia, major relapse, and postoperative ptosis have been reported.^{21,25,37,38}

KEY POINTS

- Hypertelorism is a descriptive term and not a diagnosis.
- Mild cases of hypertelorism do not require surgical treatment.
- Medial movements of the *lateral* orbital walls are required to significantly reduce the distance between the globes.
- Intracranial hypertelorism corrections are uncommonly performed procedures and may be associated with elevated surgical risks.
- Although the medial translocation of the globes is a significant surgical undertaking, the correction of associated nasal deformities is the critical finishing touch.

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26

Atypical Craniofacial Clefts

Pravin K. Patel • Henry K. Kawamoto, Jr.



he word *face* has many meanings, especially when we consider the relationship that we have with our own face. Outwardly, our face is our appearance, as we would like it to be, whether it is through the eyes of others or the image that we see reflected in a mirror. Inwardly, to some degree, our face reflects our self-worth. Our face represents our dignity and integrity; in addition, it serves as our facade and mask. This inner and outer relationship we have with our face is entwined.

Now consider when a crack runs through a face and sees itself reflected in a mirror, shattered. Is it still a face? How do we describe what remains? The child born with a craniofacial cleft is left with a sense of disfigurement and isolation. Regardless of the surgeon's attempts to restore the physical structure of the face, it is the child who reassembles and reconstructs the face from the inside out. Perhaps no other singular deformity distorts the face as much as when it is fractured by a cleft.

The meaning of the word *cleft* is apparent and easily understood when one is faced with a clinical example, such as a cleft lip or palate. However, not all defects in the apposition of junctional structures are so obviously definable as a cleft, or as easily classified. Rare or atypical clefts manifest in myriad patterns of clinical expression. This chapter includes a discussion of the developmental processes by which they may occur, an attempt to order the vast array of craniofacial clefts, and their management.

INCIDENCE OF RARE CLEFTS

The exact incidence of rare or atypical craniofacial clefts is unknown. Because of the increasing unlikelihood of finding a specific event, it is more difficult to assign a reliable number. Moreover, there is no consensus on the definition of atypical clefts. Conditions regarded as hypoplasia may

Table 26-1 Incidence of Atypical Cleft

Series	Total Number of Cleft Cases Reviewed	Number of Atypical Clefts	Ratio of Atypical to Common Clefts
Davis (1935) ⁴	946	9	10:1000
Burian (1957) ⁵	4000	97	24:1000
Fogh-Anderson (1965) ⁶	3988	48	12:1000
Popescu (1968) ⁷	1475	14	10:1000
Pitanguy (1968) ^{7a}	736	25	34:1000
Ortiz-Monasterio and Taylor (1987) ⁸	5586	174	32:1000

in reality be clefts. Treacher Collins syndrome and hemifacial microsomia should probably be included within the category of clefts. *Forme fruste* and incomplete forms of the cleft may be so minor as to go unnoticed. The incidence depends not only on the awareness of the investigator but also on the taxonomy of the facial cleft that he or she prefers.

What we are more certain of is the incidence of common clefts of the lip and palate. Cleft lip occurs at an approximate rate of 1 in 1000 live births in the white population.¹ The condition is more prevalent among the Japanese, 2.1 per 1000 births, and less common in blacks, 0.4 per 1000 births.^{2,3} It is within this context of the common cleft that the best estimate of the incidence of atypical facial cleft can be obtained. In the few published reports of a large series of patients with common clefts of the lip and palate, there is a wide range of incidences of atypical clefts⁴⁻⁸ (Table 26-1). Smaller series are even less dependable in determining the incidence of these clefts.

In a group of 937 patients with a cleft of the lip and palate, Davis⁴ found four median clefts of the lip and five oblique clefts of the face. This yielded an incidence of unusual facial clefts of 9.6 per 1000 common clefts. Burian⁵ found an incidence of 24.3 atypical facial clefts per 1000 of all cleft types in a series of 4000 clefts over a 40-year period. In a study of 3998 patients with clefts, Fogh-Andersen⁶ reported an incidence of 12 rare clefts per 1000. Of the 48 rare clefts, 15 were median clefts of the lip, 12 were transverse clefts, 8 clefts were of the nose, 7 clefts were of the scalp, 3 were oblique, and 3 were atypical clefts. Popescu,⁷ who specifically looked at transverse facial clefts alone in a group of 1475 patients, found an incidence of 9.5 per 1000. Most recently in a series of 5586 patients with clefts over a 20-year period, Ortiz-Monasterio and Taylor⁸ reported an occurrence rate of 31 atypical clefts per 1000 common clefts.

Thus the incidence of rare craniofacial clefts ranged from 9.5 to 31 per 1000 common clefts.^{9,10} How rare then is an atypical cleft? Although extrapolating these numbers to the total number of births would be erroneous, we can obtain a rough estimate, albeit only to an order of magnitude. If we accept that the incidence of the common cleft lip is 1 per 1000 live births, then an atypical cleft would be seen 100 times less frequently in the general population, and we would expect the incidence to be on the order of 1 per 100,000 births. The incidence varies not only because of its true prevalence in the population, but also with investigator awareness regarding which specific clefts were included as atypical. Within our current understanding of craniofacial clefts, the most reliable figures were those obtained by Ortiz-Monasterio and Taylor.⁸

ETIOLOGIC FACTORS AND MORPHOGENESIS OF FACIAL CLEFTS

The origins of the embryologic forces that lead to clefts are unknown. The wide spectrum of abnormalities seen may be related to slight differences in the timing, duration, or intensity of genetic and environmental insults. Differential effects in the germ layer may play a role in the phenotypic variation. Except for Treacher Collins syndrome and Goldenhar syndrome (when considered as a cleft), genetics appears to play a limited role.¹¹⁻¹³ Moreover, because atypical craniofacial clefts are rarely clinically observed or we are unable to develop a reproducible animal model, most experimental work on nongenetic factors has been performed on clefts involving the lip and palate, the most common of the facial clefts. Among the causative factors, radiation, infectious agents, metabolic imbalances, and numerous drugs and chemicals have been implicated.¹⁴⁻²⁴ Teratogenic agents in craniofacial clefting have been extensively reviewed in the literature by Kawamoto.^{9,10} Extrinsic mechanical factors from an amniotic band disruption mechanism have been suggested to explain aberrant locations of clefts not corresponding to any well-described embryologic process.²⁵⁻²⁷ Vascular disturbances have been implicated in hemifacial microsomia.²⁸⁻³²

Although it is difficult to implicate the genetic role in the vast majority of atypical clefts, the genetic basis of mandibulofacial dysostosis or Treacher Collins syndrome has more recently been established.³³ New techniques in developmental biology and molecular genetics have mapped the human genome, and with it, approximately 70 genes related to craniofacial anomalies.^{34,35} Gene linkage studies in families with clear autosomal dominance of mandibulofacial dysostosis have led the search for the causative gene to chromosome 5q31.1-q33.3.^{36,37}

As a group, craniofacial clefting is both etiologically and pathogenetically heterogeneous.^{9,10} The gaps in our understanding between causative factors, whether genetic or epigenetic, morphologic development, and its final phenotypic clinical expression are large in the vast majority of cases.³⁸ Nevertheless, an understanding of normal embryologic development forms the basis on which we understand the deviations that may occur during pathologic development.

Embryology

The structural development of the cranium and face occurs between the third and eighth weeks of gestation when fetal length measures approximately 2.0 to 31 mm.³⁹⁻⁴⁴ Fig. 26-1 is a synopsis of the relevant embryology.

Week 3: Carnegie Stages 7 Through 9, 0.4 to 2.5 mm Gestational Length

The embryo begins as a two-dimensional planar structure, and just before the third week, the stage is set for the development of the central nervous system in the area of ectoderm thickening known as the *neural plate*. A midsagittal groove appears as a result of invagination of the ectoderm centrally and simultaneous elevation of ectodermal tissue alongside the groove to form the neural folds. These folds then fuse with each other in the midline, beginning at the junction of the future brain and spinal cord, to form the neural tube. As the folds fuse, there is a separation of the neural tube from the overlying ectoderm, of which it is derived. A population of ectodermal cells adjacent to the neural fold and not included in the overlying surface (somatic) ectoderm gives rise to the formation of the neural crest. These neuroectodermal crest cells are believed to migrate widely throughout the developing embryo in a relatively cell-free-enriched extracellular matrix and differentiate into a wide array of cell and tissue types, influenced by the local environment. Most of the connective and skeletal tissues of the cranium and face are ultimately derived from derivatives of neural crest cells.

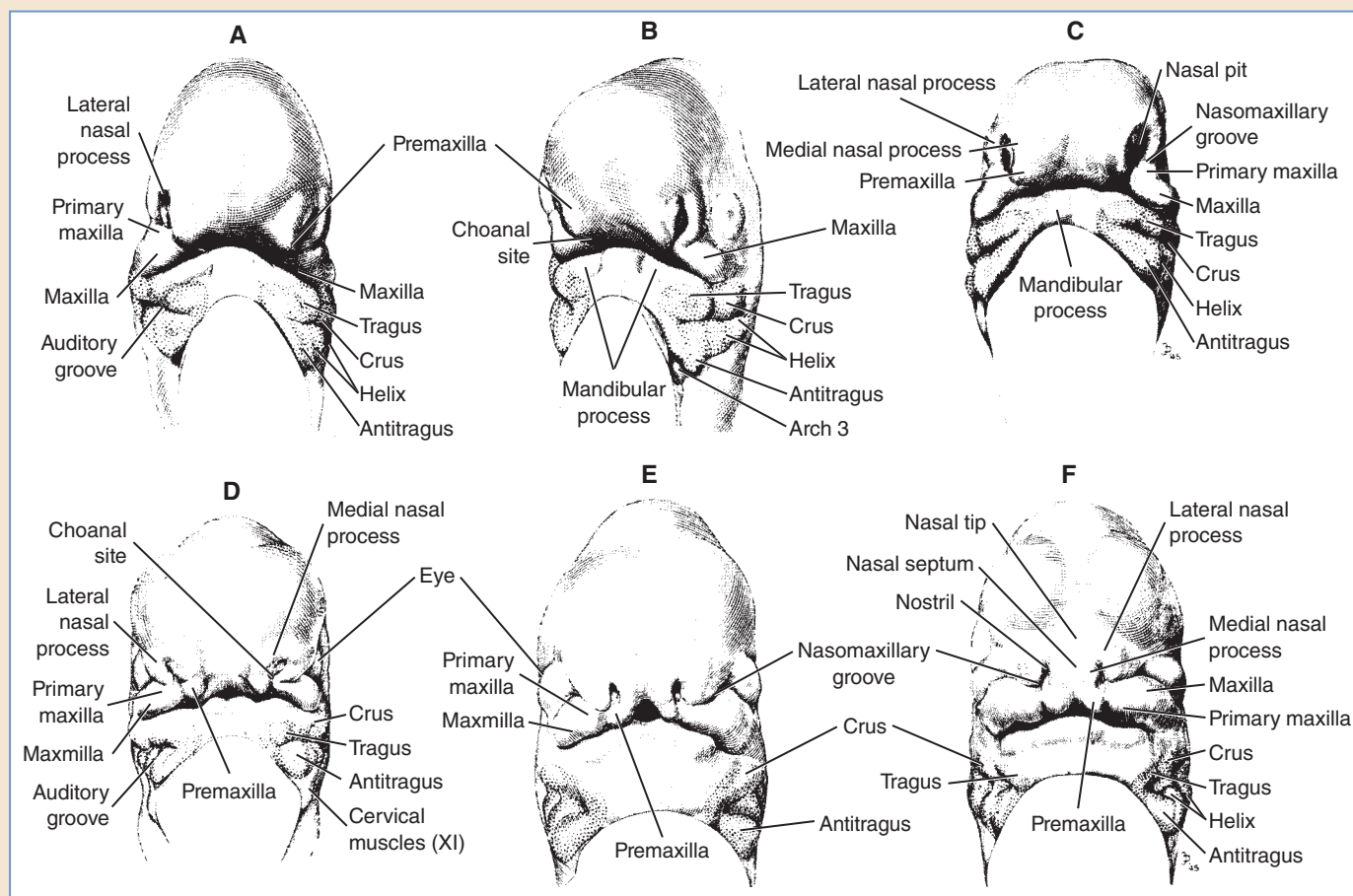


Fig. 26-1 Embryonic development of the face. A-C, Progressive development through stage 16, approximately 37 days' gestation, 8 to 11 mm. D-E, Development through stage 17, approximately 41 days, 11 to 14 mm.

The first morphologic evidence of the optic primordia is seen as a thickened area with a shallow sulcus on the lateral forebrain of the neural tube in the region of the future diencephalon. During the week, the optic sulcus deepens, and the surrounding walls form an evagination, which at first is in contact with the overlying ectoderm. Neural crest cells intervene between the optic vesicle and overlying ectoderm and give rise to specialized neuroectodermal elements of the eye and adnexa.

As the neural tube fuses cranially, closing off the anterior neuropore, a mesencephalic flexure occurs and the major divisions of the brain—*prosencephalon* (forebrain), *mesencephalon* (midbrain), and *rhombencephalon* (hindbrain)—can begin to be distinguished. By the end of the third week, further growth of the cephalic portion of the neural tube gives rise to the frontonasal process and branchial arches—the first stages of a recognizable face.

Week 4: Carnegie Stages 10 Through 13, 2.0 to 6.0 mm Gestational Length

The recognizable face begins its development from five primordia that surround a central area of depression, the oral pit or stomodeum. These primordia are a single cranially located fron-

tonasal process, two bilaterally located maxillary processes, and mandibular processes. The maxillary and mandibular processes are derived from the first branchial arch. These processes or masses grow differentially, and by obliterating the ectodermal plates or grooves between them, eventually give rise to the features of the face. The mandibular processes are the first to merge with each other at the midline and will eventually give rise to the mandible and lower part of the face and tongue.

During the fourth week the primitive trisegmented brain further subdivides. The prosencephalon (forebrain) divides into the telencephalon (endbrain) with prominent lateral domes (cerebral hemispheres) and the diencephalon, which gives rise to the optic vesicles. The mesencephalon remains in flexed cephalic flexure and undivided. The rhombencephalon divides into the metencephalon (cerebellum and pons) and the myelencephalon (medulla). By the end of the fourth week, the fundamental organization of the future brain is clearly identifiable.

The ectoderm overlying the optic vesicle of the developing eye thickens to form the lens placode. During the week, the optic vesicle deepens to form a goblet-shaped, double-layered optic cup as its stem narrows to form the optic stalk in continuity with the diencephalon. The frontonasal process intervenes between the two lateral optic cups, and at this stage the developing eyes are nearly 180 degrees apart.^{43,45}

Week 5: Carnegie Stages 14 and 15, 5.0 to 9.0 mm Gestational Length

Toward the end of the fourth week, nasal placodes develop bilaterally at the inferolateral corners of the frontonasal process. As the region surrounding the nasal placodes develops into medial and lateral nasal processes, the deepening nasal placodes are transformed into nasal pits, which in time form the anterior nares. With continued ectodermal development, the nasal placodes become olfactory grooves that at this stage are in continuity with the developing oral cavity. The maxillary processes continue to enlarge, encroaching on the stomodeum, to form a primitive oral cavity.

As the optic stalk narrows further to define the primitive optic nerve, retinal pigment appears in the external layer of the optic cup, and the inner layer begins to differentiate into the neural elements of the retina. The primitive vitreous body develops in the intervening space as the lens placode invaginates toward the optic cup to form a lens vesicle detaching itself from the overlying ectoderm.^{43,45}

As the features of the face form, the development of the embryonic skull begins. The mesenchyme surrounding the developing primitive brain condenses to form a mesenchymal capsule, the *desmocranium*. At about the fifth week, the base thickens, forming the cartilaginous cranial base through neuroectodermal condensation of the mesenchyme. In contrast, the calvarium develops by direct membranous ossification without a cartilaginous precursor later in the embryonic period.⁴⁶

Week 6: Carnegie Stages 17 Through 19, 8.0 to 14 mm Gestational Length

The medial nasal processes approach each other to form a single globular process that in time will give rise to the nasal tip, columella, prolabium, frenulum, and primary palate. As this occurs, the frontonasal process collapses inward to form the nasal septum. Continued growth of the maxillary mass below the optic vesicles allows fusion with the lateral nasal process. As fusion occurs, a sinking troughlike epithelial groove demarcates the nasooptic furrow connecting the conjunctival lacrimal sac with the lateral nasal wall. The epithelial groove fuses as a solid epithelial cord and eventually canalizes later in fetal development to become the nasolacrimal duct.⁴⁷ Toward the end of week 6, the maxillary process fuses with the medial nasal fold of the globular process, forming a true nostril as it gives rise to the lateral lip element (Fig. 26-2). However, posterior to this anterior fusion of the maxillary process to the nasal processes, the developing nasal floor is open to the oral cavity. Within the primitive stomodeum, lateral palatine processes develop from

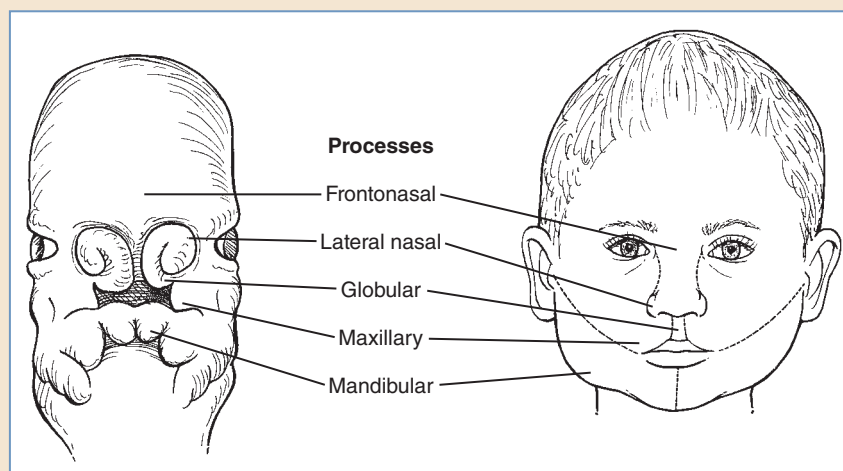


Fig. 26-2 Contributions of the embryonic processes to the final emergence of the face.

the medial edges from the maxillary process to give rise to the secondary palate. At this stage, the developing tongue nearly completely fills the oral nasal cavity and reaches the nasal septum.

Simultaneously, the eye continues its development as neurocrest mesoectodermal cells at the lateral aspect of the lens differentiate into the early cornea and sclera. The cavity of the optic stalk is obliterated as nerve fibers from the retina grow back to reach the brain to form the optic nerve. The overlying surface ectoderm develops subtle horizontal folds with a mesenchymal core above and below the developing cornea that represent the beginning of the upper and lower eyelids.^{43,45,48}

During this sixth week, the external ear develops from six mesenchymal swellings (hillocks) that surround the first branchial cleft. The first three hillocks arise from the mandibular arch (first branchial arch) and the second three arise from the hyoid arch (second branchial arch) (Fig. 26-3). From ventral to dorsum, the hillocks of the first arch become the tragus, helix, and cymba concha; the hillocks on the second arch, the antitragus, antihelix, and concha. The branchial cleft lengthens to form the primordium of the external auditory canal. As the face develops, the auricle is gradually repositioned to a more lateral cephalic position from the original location low on the side of the neck.⁴⁵

Week 8: Carnegie Stages 20 Through 23, 18 to 31 mm Gestational Length

By the eighth week, the face has acquired a more human appearance, with complete closure of the lower facial fissures or grooves. The primitive upper and lower jaws are formed with complete fusion of the maxillary and mandibular processes. The upper lip and lower nasal regions are better defined. As it lengthens vertically, the frontonasal process continues to collapse horizontally, forming a transverse furrow at the nasal bridge. As the frontocranial region loses its overhang, the eyes begin to take a more medial position. The anterior rotation of the eyes continues throughout the remainder of the gestational period and well into childhood (Fig. 26-4). The upper and lower lids continue to develop and assume a more almond shape as they begin to fuse with each other (separation of the lids does not occur until weeks 25 to 26). The lined ectodermal-derived

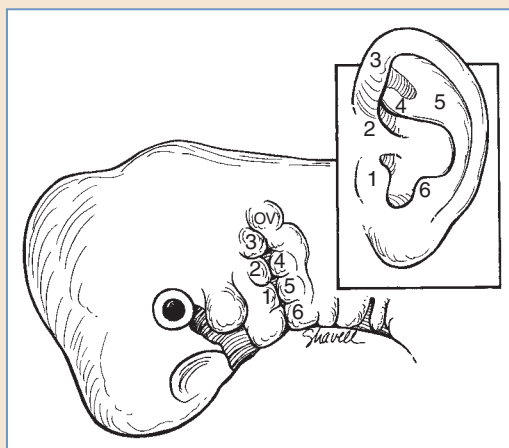


Fig. 26-3 Development of the external ear illustrating the contribution of the hillocks from the first (1-3) and second branchial arches (4-6).

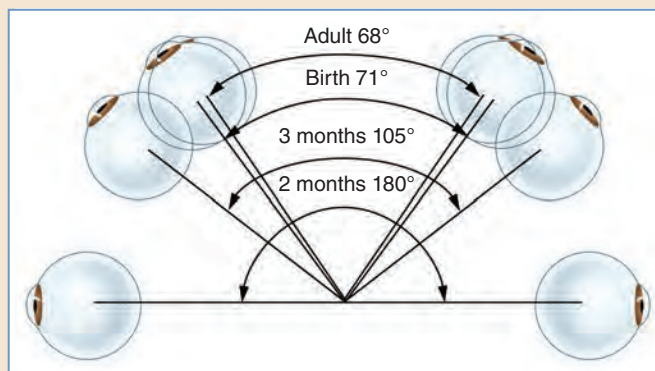


Fig. 26-4 Progressive medial rotation of the orbit and ocular globe through gestation and into adulthood.

epithelium between the fused eyelids and the cornea develops into the conjunctival sac and from the superolateral angles, the lacrimal glands.⁴³

The development of the skull continues, with ossification centers arising within the *mesenchyme* (membranous ossification) adjacent to the developing brain to form the flat bones of the calvarial vault, the paired frontal and parietal bones (Fig. 26-5). In contrast, the primordial elements of the chondrocranium begin to undergo endochondral ossification to form the bones of the *basicranium* (sphenoid, petrous part of the temporal and occipital bones) and nasal ethmoid significantly later in fetal life. The sphenoethmoidal and sphenooccipital synchondroses and septal cartilage persist to allow the forward growth of the cranial base and simultaneously to allow the inward rotation of the developing orbital complex.^{46,49,50}

In the face, small nasal bones appear above the nasal capsule, the cartilaginous model. On the lateral surface of the nasal capsule, premaxillary and maxillary ossification centers appear. Posteriolateral to the developing maxillary bone, ossification centers arise within the zygomatic

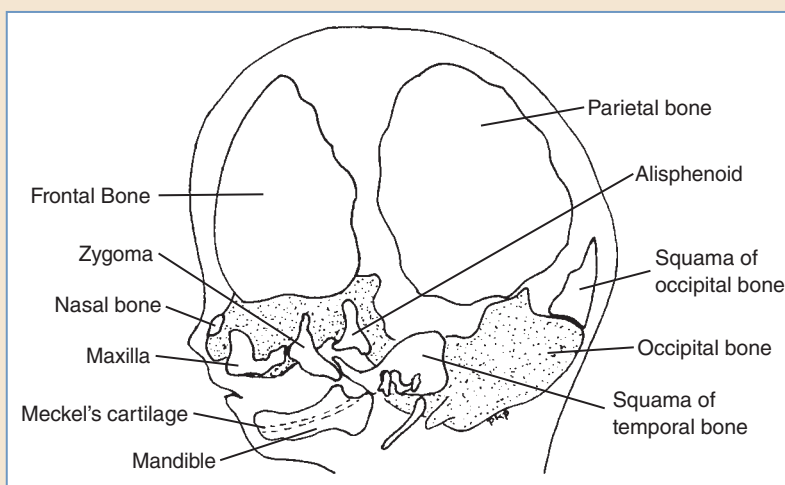


Fig. 26-5 Development of the craniofacial skeleton. Mesenchymal formation of the facial bones derived from neural crest cell contribution (*stippled areas*).

and squamosal portion of the temporal region. The mesenchyme encompassing Meckel's cartilage undergoes membranous ossification to form the body of the mandible. In time, Meckel's cartilage disappears and persists as the incus and malleolus of the middle ear. The onset of bone formation marks the end of the embryonic period, and the developing fetus is generally much more resistant to teratogenic insults.⁴⁴

Within this short period of 5 weeks, when the mother may be unaware of her own pregnancy, the face of her child has emerged. Any error that occurs in these tightly sequenced events of early facial embryogenesis can have significant consequences. By whatever causative factor, failure of the lateral maxillary process to fuse with the central globular process would be expected to result in a unilateral cleft of the upper lip. Similarly, the median cleft can be explained by incomplete merging of the median nasal processes and an oblique facial cleft by persistence of the groove between the maxillary and lateral nasal processes (medial canthal region to the ala of the nose).

Although it is understandable that some facial clefts occur when there is a lack of fusion of the various primordial facial processes, others are not readily explainable in such terms. A cleft that begins lateral to the philtrum and extends to the midportion of the lower eyelid is difficult to explain. As such, these atypical clefts, which are not obviously demarcated in the developing face, were considered ruptures of the embryonic tissue by external forces.

In the literature, differences of opinion exist regarding what fusion itself means in embryogenesis and what role cell proliferation, cell differentiation, cell degeneration, and the extracellular matrix play in morphogenesis.

MORPHOGENESIS OF FACIAL CLEFTING

Historically, two theories of facial clefting have been described: (1) the classic theory of fusion by Dursy⁵¹ and His⁵² and (2) the theory of mesodermal migration by Veau and Politzer,⁵³ Warbrick,⁵⁴ Stark,⁵⁵ and others. More recently, theories proposed by Johnston⁵⁶ and Vermeij-Keers et al⁵⁷ offered possible explanations of some of the more rare defects clinically seen.

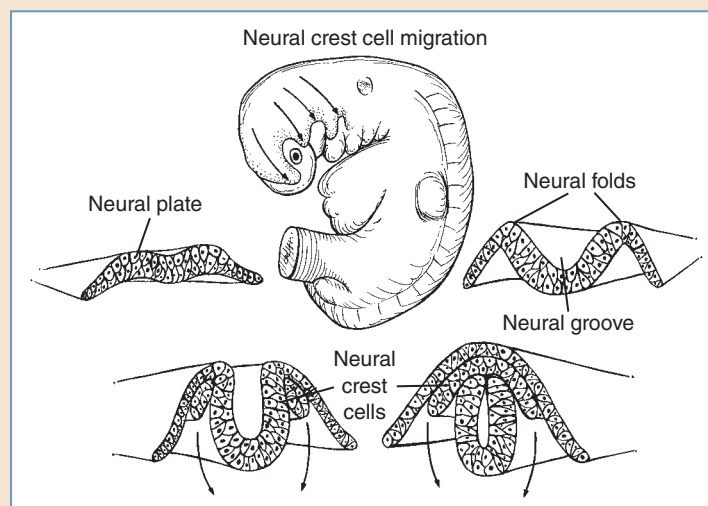


Fig. 26-6 Development of the neural plate and origin of neural crest cells. Neural crest cells are believed to play a significant role, contributing to the mesenchyme of the craniofacial skeleton and connective tissue.

Proposed by Dursy in 1869⁵¹ and by His in 1892,⁵² the classic theory suggests that clefts develop because of a lack of fusion of the normally merging facial processes. After the free, bud-like ends of the facial processes establish ectodermal contact, mesodermal penetration occurs to complete the union. The upper lip is formed by the union of the frontonasal process centrally with the lateral maxillary processes. Disruption of the sequence—fusion followed by mesodermal penetration—leads to the various clefts of the upper lip.^{51,52}

In contrast to the classic theory, the theory of mesodermal migration emerged in the first half of the twentieth century. Its supporters believed that the free ends of the facial processes did not exist. Instead, the developing embryonic face was composed of a continuous bilamellar ectodermal membrane, with epithelial seams demarcating the various processes of the face. Within this bilamellar structure, mesenchyme migrates and penetrates to smooth out the seams.⁵⁸ Thus clefting seems to be related not to failure of fusion but to failure of mesenchymal migration. The facial processes as such do not fuse but rather merge.^{53,54,59}

Weston⁶⁰ and Johnston⁶¹ further refined the concept of mesodermal migration when they showed the importance of neural crest cells in the development of the mesodermal component of the craniofacial form. Their work identified a specialized group of cells, the *neuroectodermal cells*, arising from the dorsal lateral ectoderm of the neural fold to migrate extensively to their eventual destination along hyaluronate-rich, fibronectin-lined extracellular pathways.⁶² The cells play a significant role in contributing to the mesenchyme responsible for skeletal and connective tissue of the face (Fig. 26-6). The cells from the differing levels of the neural fold eventually populate specific regions: (1) cells from the prosencephalon and cranial mesencephalic levels of the neural tube populate the region of the frontonasal prominence, and (2) cells from the caudal mesencephalic and upper rhombencephalic region populate the primitive maxilla and developing mandible. This cranial-to-caudal sequence is preserved.^{63,64} If the migration is incomplete or insufficient in number or undergoes abnormal differentiation, various malformations then result. The severity depends on the stage at which the failure occurs. If mesodermal penetration and

differentiation by the neuroectodermal cells fail to occur, a facial cleft manifests as the epithelial seam breaks down.^{56,65}

More recently, studies by Vermeij-Keers et al⁵⁷ and others have shown migration of neural crest cells alone may not account for all varieties of facial clefting.⁶⁶ There is disagreement in the literature regarding whether the neural crest cells migrate or are developmental rests as the neural plate is transformed three dimensionally.^{56,62,64-68} Johnston's conclusions⁵⁶ of neural crest migration were based on nonmammalian vertebrate embryos. In contrast, studies based on murine embryos have failed to confirm the migration of neural crest cells relative to their surrounding mesenchyme. Instead, it appears that the neural crest cells are not moving alone through an extracellular matrix but are part of an integrated coordinated set of tissue displacements.^{63,67} Facial clefts then are not necessarily related to failure of migration but rather to failure of local differentiation of the neuroectoderm and mesenchyme.

Vermeij-Keers et al⁵⁷ identified four primary sites in early embryologic development that could lead to cleft formation between:

1. The junction of the medial and lateral nasal processes and maxillary process
2. The lateral nasal and maxillary processes
3. The maxillary and mandibular processes
4. The palatine processes

These sites are then the origins of true clefts, because they are associated with the normal planes of embryologic processes. All the defects arising at the site of the epithelial plate show an ectodermal defect. Closure of the ectoderm of the face occurs by the 17 mm crown-rump (CR) length stage, which is sometime within the seventh gestational week. Thereafter, within each facial element, differentiation of the mesenchyme into facial muscles and supporting bone and cartilaginous support occurs. Ossification centers arise to complete bone formation. If the ossification centers fail, then pseudoclefts develop. These are not true clefts, and as such, the terms *secondary cleft* or *dysplasia* were suggested by Vermeij-Keers et al⁵⁷ and van der Meulen et al.⁶⁹ Errors in early facial development, before the 17 mm CR stage, lead to true clefts; errors in the late embryonic stage, after the 17 mm CR stage, result in dysplasia or a pseudocleft. The location of the primary or true clefts is well defined. The clefts of the later embryonic stage of development depend not only on the well-defined set of membranous bones but also on the number and location of the ossification centers within each of the elements.⁴⁹ The atypical clefts are then believed to be accounted for by the variability in the number of ossification centers within each of the developing bony plates.⁶⁶ Sulik⁷⁰ has suggested that when human embryos at 5 and 6 weeks' gestation are carefully examined with scanning electron microscopy, the more familiar facial processes (nasal, maxillary, and mandibular) are composed of smaller units, representing possible growth centers. The boundaries between the growth centers can be correlated with the potential sites of atypical facial clefts⁷⁰ (Fig. 26-7).

Early in the development, when the brain and facial form are intimately connected, variable degrees of deficiency of the anterior neural plate lead to a series of malformations termed *holoprosencephaly*.⁷¹⁻⁷⁴ The name derives from when there is incomplete or complete failure of the anterior portion of the neural tube to form the cerebral hemispheres, resulting in a single (holo) forebrain (*prosencephalon*)⁷⁵ (Fig. 26-8). Such early failures before the third gestational week of embryogenesis also are associated with various monstrous facial dysmorphisms. If the prosencephalon and frontonasal process fail to develop, cyclopia, ethmocephaly, and cebocephaly can occur. In *cyclopia*, the most extreme form, there is a single median eye or closely approximated paired ocular structures (*synophthalmia*) in a single orbit associated with arhinia and a rudimentary proboscis above the ocular field. In *ethmocephaly*, the hypoteloric eyes are structurally separate with an intervening proboscis. In *cebocephaly*, the ocular hypotelorism is to a lesser degree, and the proboscis is below the ocular field and more closely resembles a rudimentary nose with a blind single nostril.⁷⁶

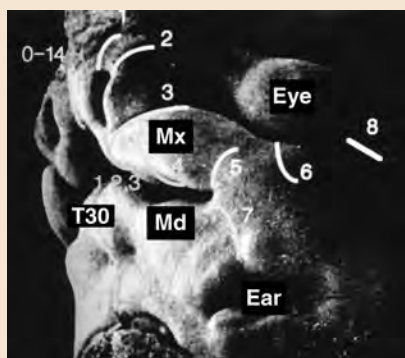


Fig. 26-7 Embryonic lines superimposed on an embryo at 6 weeks' gestation. The potential clefts are numbered according to Tessier's classification from 0 to 14, along with the mandibular midline Tessier 30 cleft. Although not formally identified in Tessier's classification, the paramedian cleft is noted as Tessier 29/28 at the junction of the two mandibular prominences. (*Md*, Mandible; *Mx*, maxilla; *T30*, Tessier 30 cleft.)

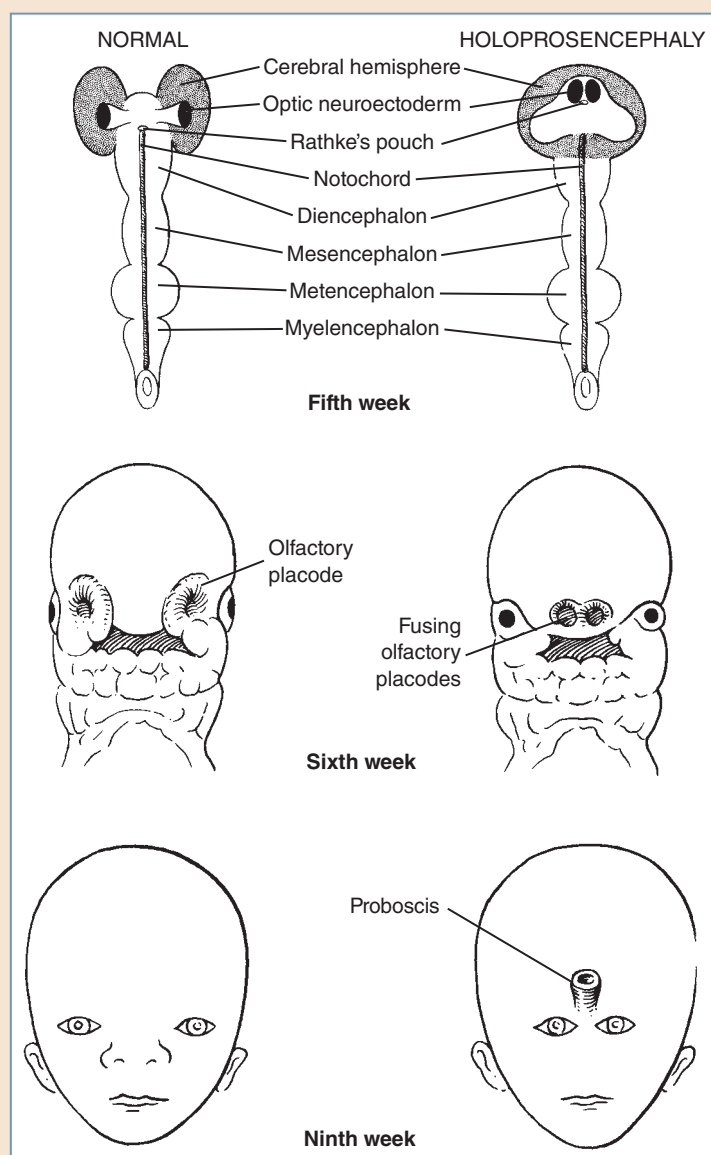


Fig. 26-8 Holoprosencephaly. The prosencephalon fails to cleave sagittally into cerebral hemispheres. Convergence of the optic and olfactory placodes results in failure of development of the embryonic brain and frontonasal process.

In contrast, median facial defects characterized by orbital hypertelorism are believed to occur late, after the 17 mm CR stage. The ocular complex remains in the lateral position as a result of the intervening frontonasal process arrested in the low embryonic position. Normally, the internasal groove disappears with the forward growth (frontocaudal direction) of the nasal septum and simultaneously the distance between the ocular complexes decreases. When this fails, orbital hypertelorism occurs, wherein both distances between the medial orbital and lateral orbital walls are increased.⁷⁷

There is an extensive body of literature in which the typical clefts and various atypical clefts can be related to fundamental embryologic processes based on more recent concepts of neuromeric organization. The embryonic brain and spinal cord are divided into functional segments along the neuraxis that resemble a string of pearls. These units are called *neuromeres*. The forebrain, or prosencephalon, contains six prosomeres. Prosomeres 1 through 3 and 4 through 6 constitute the primitive diencephalon and telencephalon, respectively. The midbrain, or mesencephalon, is derived from *mesomeres* 1 and 2. *Rhombomeres* 0 through 11 constitute the primitive hindbrain, or *rhombencephalon*. Each neuromere proceeding caudally from the hindbrain is flanked by a *somitomere*, which subsequently differentiates into formed structures. Pertinent to facial anatomy, the first seven somitomeres form the cranial base dorsally and the first three pharyngeal arches ventrally. The fourth through the sixth pharyngeal arches are formed by the ventral mesoderm of somitomeres 8 through 10. The morphology of the face is produced by the assembly of embryonic fields according to a genetic program. Fields represent true neuroangiosomes that move analogously to tectonic plates, whereas processes merely represent the topography of the embryonic face. Clefting is believed to result from the mechanical disruption of programmed field interactions. Carstens⁷⁸ details the contemporary view of head and neck embryology, which is relevant to the plastic surgeon who treats congenital deformities.⁷⁹

CLASSIFYING THE RARE CRANIOFACIAL CLEFT

Classification of Median Facial Anomalies

The *median cerebrofacial malformations* are developmental anomalies of the midline brain and facial structures. Unlike the oblique facial clefts, the median facial defects can more readily be understood on an embryologic basis and consequently are easier to classify. As a group of midline deformities, these can be broadly subdivided into (1) those involving a deficiency in tissue and (2) those involving either an excess or near-normal amount of intervening tissue. However, extremes of malformation in both groups represent opposite ends of a continuous spectrum of median facial anomalies.

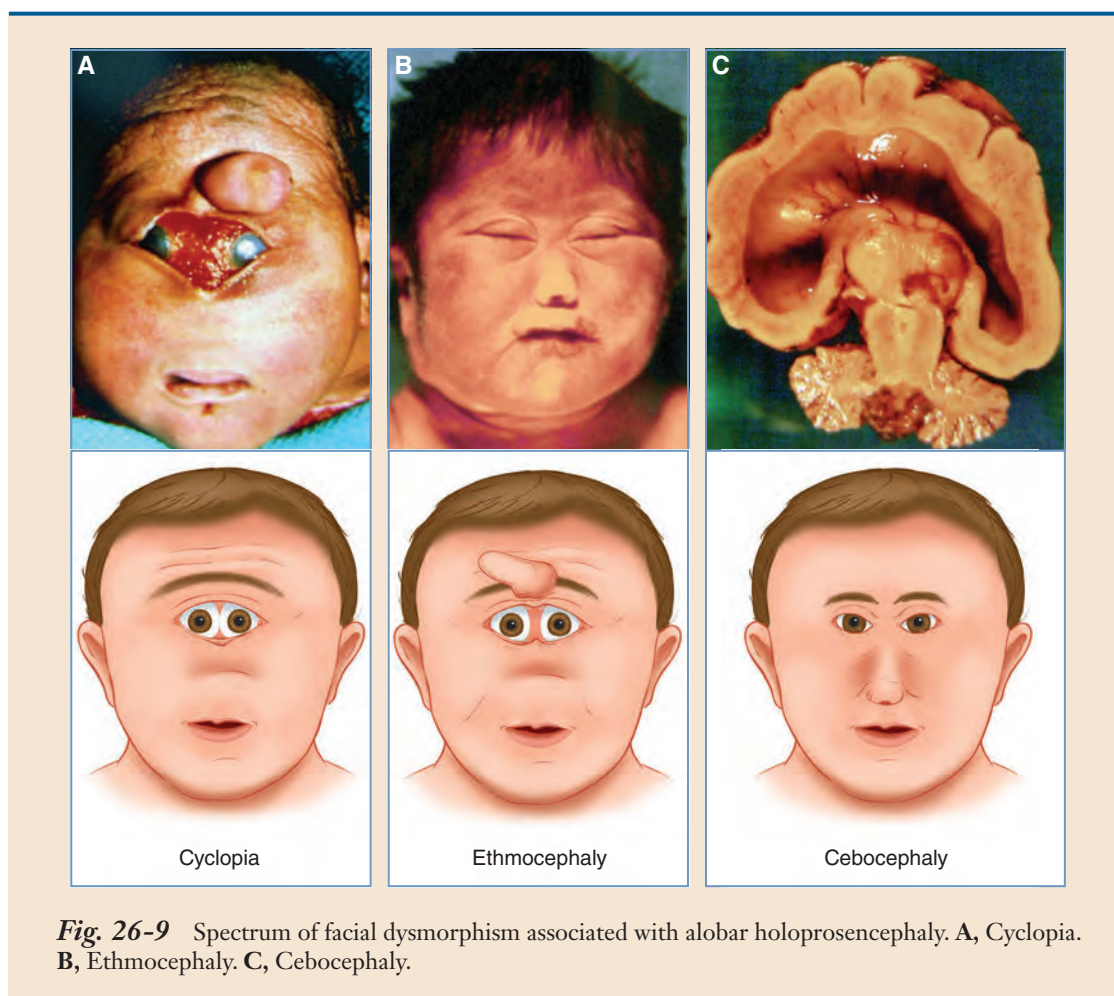
Tissue-Deficient Median Cerebrofacial Dysmorphogenesis

In an attempt to categorize cases of median facial anomalies in which there is deficient central cerebral/facial tissue, in 1963 DeMyer and Zeman⁷⁵ proposed the term *holoprosencephaly*. The prosencephalon remains “holistic,” failing to cleave sagittally into the cerebral hemispheres, transversely into the telencephalon and diencephalon, or horizontally into olfactory and optic bulbs. These cases can be graded to the degree of severity as to lobar, semilobar, and alobar holoprosencephaly (Fig. 26-9). Embryologically, an intimate relationship exists between the developing midfacial structures and forebrain, and as such, the degree of facial disorganization reflects an equally severe anomaly of the brain.^{71,80} The varying degrees of facial dysmorphism associated with the severity of holoprosencephaly have been grouped into five major types^{72,77,80} (Table 26-2):

- I. Cycopia
- II. Ethmocephaly
- III. Cebocephaly
- IV. Hypotelorism with median cleft lip
- V. Hypotelorism with bilateral cleft lip

Table 26-2 Holoprosencephaly

Type/Group	Facial Morphology	Brain Morphology
I Cyclopia	Monophthalmia, synophthalmia, or anophthalmia Proboscis (single/double), arhinia	Alobar
II Ethmocephaly	Extreme hypotelorism Proboscis (single/double), arhinia	Alobar
III Cebocephaly	Hypotelorism Proboscis-type nose (blind end/single nostril)	Alobar
IV	Hypotelorism Flat nose Median cleft lip	Alobar (usually)
V	Hypotelorism Flat nose Bilateral cleft lip	Semilobar or lobar



The degree of correlation between the brain and face is not absolute. The predictive correlation is only about 80%.^{81,82} For the clinician deciding which infant may be a candidate for reconstruction, the distinction between the groups becomes clinically relevant. Infants who fall into types I, II, or III, in which the brain morphology is alobar, rarely survive beyond the first year of life and are not candidates for reconstruction. The difficulty lies in determining which infants in the type IV and V categories should be offered reconstructive surgery. The classification scheme of DeMyer et al,⁸⁰ which was later modified by Cohen et al,⁷² Cohen and Sulik,⁷⁶ and Cohen⁸³ is based primarily on the cleavage of the brain. There remains a wide spectrum of midfacial hypoplasia and false median clefts that can occur in the absence of any significant brain pathology.⁸⁴⁻⁸⁹ These last two categories are more usefully further stratified based on prognostic and functional criteria in the proposed reclassification of Elias et al⁹⁰ (Table 26-3).

In the revised scheme, *types I* through *III* remain unchanged, because they all reflect alobar holoprosencephaly. *Type IV* as a transition includes either alobar or semilobar holoprosencephaly, and *Type V* includes lobar holoprosencephaly. *Type VI* would be added to represent normal brain development, and yet facial dysmorphogenesis is consistent with the characteristics of this group of deformities. *Type VI* can be further subdivided based on the presence or absence of a false median cleft lip. Despite similarities in dysmorphic facial appearance, *type VI* cannot be considered a true holoprosencephaly, because the brain is anatomically normal. Our experience has been that infants with revised *types V* and *VI* facial deformity should be given the opportunity of surgical reconstruction. Although infants in type V are invariably mentally deficient, their potential for life expectancy justifies early repair of the lip and palate. Similarly, infants in *group VIa* should undergo early lip and palate repair. Unlike *type V* infants, a further major craniofacial procedure should be contemplated with *type VI* deformities, with the understanding that subtle neurologic deficits may exist.⁹⁰

Table 26-3 Median Cerebrofacial Dysmorphogenesis

Type/Group	Facial Morphology	Brain Morphology		Operative Procedure
I Cyclopia	Monophthalmia, synophthalmia, anophthalmia Proboscis (single/double), arhinia	Alobar	HOLOPROSENCEPHALY	None
II Ethmocephaly	Extreme hypotelorism Proboscis (single/double), arhinia	Alobar		None
III Cebocephaly	Hypotelorism Proboscis type nose (blind end/single nostril)	Alobar		None
IV	Hypotelorism Flat nose False median cleft lip	Alobar/semilobar		None
V	Hypotelorism Flat nose False median cleft lip	Lobar		Lip/palate repair only
VI	Hypotelorism Flat nose	Normal		Lip/palate repair LeFort III
VIa	False median cleft lip			Facial bipartition
VIb	No false median cleft lip			

Tissue Excess or Near-Normal Median Cerebrofacial Dysmorphogenesis

DeMyer⁹¹ proposed the term *median cleft face syndrome* to describe this collection of median facial anomalies characterized by near-normal or excess tissue. The dysmorphic features can vary from a subtle midline notch of the upper lip to an obvious bifidity of the nose, and in more severe cases hypertelorism (Fig. 26-10). Characteristic features of the syndrome include:

- Orbital hypertelorism
- V-shaped frontal hairline
- Bifid cranium
- Median cleft of the upper lip
- Median cleft of the premaxilla
- Median cleft of the palate
- Primary telecanthus

Unlike holoprosencephaly, there is little predictive value between the face and underlying brain. The likelihood of mental retardation increases in patients with severe hypertelorism or if it is combined with extracephalic deformities.⁹¹

Instead, Sedano et al⁹² preferred the term *frontonasal dysplasia* and first suggested that this group of midline defects likely represents the opposite end of the spectrum from holoprosencephaly. Their preference for the term frontonasal dysplasia is objectionable. The term neither accurately describes the median nature of these defects nor does it seemingly include median labial-palatal defects. The term is vague as it has been used in the literature. Moreover, it has included nasal alar clefts that are not true midline defects. DeMyer's⁹¹ more preferable term emphasizes the median nature of this group of anomalies but is also objectionable, because it implies a syndromic feature to these defects. We prefer instead the term *median cleft face dysmorphism* to describe this category.

DeMyer's inclusion of primary telecanthus⁹¹ as a characteristic should also be dropped, because it is not a true midline defect. *Primary telecanthus* more accurately describes the lateral displacement of the medial canthi as seen in blepharophimosis. *Hypertelorism* emphasizes the skeletal deformity, in which there is an increased distance between the medial orbital walls. Although hypertelorism is a characteristic physical finding in this group of midline facial clefts, paramedian clefts can also result in hypertelorism.^{9,10}



Fig. 26-10 Median cleft face dysmorphism with central tissue excess represents the opposite end of the spectrum from holoprosencephaly.

Classification of Oblique Clefts

For decades the task of classifying rare oblique craniofacial clefts has posed a challenge. Since the numerous early attempts at classification by Morian in 1887,⁹³ Sanvenero-Rosselli in 1953,⁹⁴ Burian in 1960,⁹⁵ Lund in 1966,⁹⁶ the rare or atypical clefts have seemed to defy any simple and obvious classification. The clefts are rare, the descriptions in the literature are incomplete, and the definitions are not necessarily agreed on. In more recent years, several different classifications have been proposed, but none have gained absolute universal acceptance by all investigators. The proposed classification schemes have been based purely on either observed anatomic lines without regard to any underlying causative principles (for example, by Boo-Chai,⁹⁷ Tessier,⁹⁸ and Harkins et al⁹⁹) or on an understanding of embryologic studies (for example, Karfik¹⁰⁰ and Van der Meulen et al⁶⁹). The basic scientist prefers an embryologic classification, whereas the clinician prefers simplicity with clinical applicability.

In 1962 the American Association of Cleft Palate Rehabilitation⁹⁷ classified the rare facial clefts into four major groups: (1) mandibular process clefts, (2) nasoocular clefts, (3) oroocular clefts, and (4) oroaural clefts (Fig. 26-11). The oroocular clefts were further subdivided into oral medial canthus and oral lateral canthus clefts with a temporal extension. The midfacial defects and clefts related to Treacher Collins syndrome were omitted in this classification. Moreover, because it was based only on surface anatomy, this simple classification scheme failed to consider the underlying skeletal pathology.

In 1970 Boo-Chai,⁹⁷ who recognized Morian's initial observations from 1887,⁹³ subdivided the oroocular clefts more specifically into two types: *Type I clefts* began just lateral to the philtral ridge and continued in a cephalic direction to end in the region of the medial canthus. Its skeletal counterpart began between the lateral incisor and canine, coursing medial to the inferior orbital foramen, and on to the inferior medial aspect of the orbit. In contrast, *type II clefts* began more laterally near the oral commissure and ended either at the midportion of the eyelid or lateral

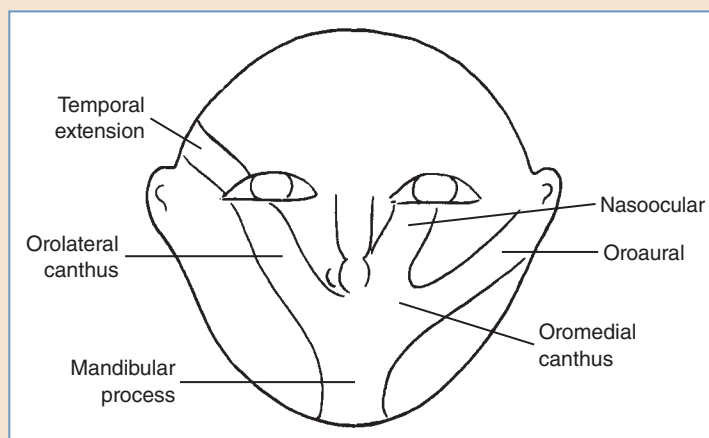


Fig. 26-11 Classification scheme endorsed by the American Association of Cleft Palate Rehabilitation in 1962. The clefts were broadly classified into one of four major categories: mandibular, nasoocular, oroocular, and oroaural.

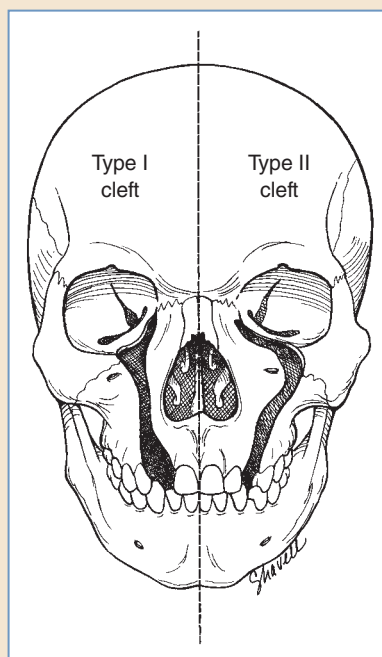


Fig. 26-12 Classification scheme proposed by Boo-Chai in 1970 for oroocular clefts. The clefts are divided according to whether they course medial (type I) or lateral (type II) to the infraorbital nerve.

canthus. The skeletal cleft began in the premolar region, coursed lateral to the inferior orbital foramen, and terminated at the inferior lateral orbit (Fig. 26-12).

In contrast to the above attempts at a phenomenologic classification of clefts, in 1966 Karfik¹⁰⁰ presented a classification based on embryologic and morphologic grounds (Box 26-1). The cleft malformations were divided into five major groups¹⁰⁰:

1. Rhinencephalic disorders
2. Branchiogenic disorders
3. Ophthalmoorbital disorders
4. Craniocephalic disorders
5. Atypical facial disorders

Unfortunately, the oroocular clefts could not be clearly categorized. Group A includes not only the nasoocular clefts but also the oroocular clefts of type I described by Boo-Chai⁹⁷ and Morian,⁹³ which do not have nasal involvement and start lateral to the common cleft lip. Type II oroocular clefts were grouped in group E under atypical facial disorders, along with facial asymmetries, neoplasms, and others that could not be explained by an embryologic seam. Moreover, Apert and Crouzon syndromes are included in group D, which today would be considered dysostosis rather than a clefting phenomenon.

Box 26-1 Facial Cleft According to Karfik**Group A: Rhinencephalic Disorders*****A1, Axial***

Prolapse: Meningocele, glioma, dermoid cyst, teratoma

Clefts: Medial nasal (double nose), median cleft of the upper lip and premaxilla

Defects: Coloboma of nostril, partial nose, total nose, septal, atresia nasi

A2, Paraaxial Clefts

Coloboma, iris or palpebral, total or partial paraaxial cleft lip, typical, lacrimal duct dystopia

Group B: Branchiogenic Disorders***B1, Lateral Otocephalic***

Clefts: Macrostomia, lateral cervical fistula

Dysostosis: Mandibular (for example, Pierre Robin sequence), mandibulofacial (for example, Treacher Collins syndrome)

Defects: Partial or total auricular, atresia

B2, Median Axial Clefts

Tongue, lower lip, mandible, fissura colli medialis, fissura thoracis medialis

Group C: Ophthalmoorbital Disorders

Malformations: Eyeball: microphthalmia, anophthalmia; lids: Blepharophimosis, epicanthus

Defects: Orbital

Group D: Craniocephalic Disorders

Malformations: Head and face (for example, Apert syndrome, Crouzon disease)

Defects: Scalp, skull

Group E: Atypical Facial Disorders

Oblique facial cleft, parasitic dysembryoma, hemifacial atrophy, hyperplasia, congenital neoplasm, teratoma

Subsequently, in 1983, Vermeij-Keers et al⁵⁷ proposed a classification scheme based on the embryologic work of van der Meulen⁶⁹ (Box 26-2). As previously discussed, van der Meulen et al⁶⁹ showed that the underlying pathologic findings of the malformations can be interpreted as developmental arrests that occurred either before the fusion of the facial processes (17 mm CR stage of the embryo) or after—during the differentiation phase. The early defects result in true clefts, or primary clefts, along embryologic processes. The late defects, caused by errors in differentiation and fusion of ossification centers, result in pseudoclefts, or secondary clefts.⁶⁶

Objecting to the use of the term *cleft*, Vermeij-Keers et al⁵⁷ proposed that the more general term *dysplasia* would be more applicable, because the defects seen do not necessarily represent obvious clefts, depending on the severity of involvement. Both true clefts (for example, cleft lip and palate) and pseudoclefts (Treacher Collins syndrome and hemifacial microsomia) would be regarded as dysplasia in the classification scheme.⁶⁹ Moreover, the dysplasia should be properly identified by the area of the facial processes involved rather than assigned a random numeric classification. Furthermore, a distinction should be made between malformations of the face and cranium alone (*craniofacial dysplasia*) and craniofacial defects involving the brain (*cerebrofacial dysplasia*), which occur earlier in the embryologic development. Craniofacial dysplasia is further

Box 26-2 Classification of Craniofacial Malformations According to van der Meulen et al

- I. Cerebrocranial Dysplasias
- II. Cerebrofacial Dysplasias
 - A. Rhinencephalic dysplasias
 - B. Oculoorbital dysplasias
- III. Craniofacial Dysplasias
 - A. With clefting
 - 1. Lateronasomaxillary cleft
 - 2. Medionasomaxillary cleft
 - 3. Intermaxillary cleft
 - 4. Maxillomandibular cleft
 - B. With dysostosis
 - 1. Sphenoidal
 - 2. Sphenofrontal
 - 3. Frontal
 - 4. Frontofrontal
 - 5. Frontoethmoidal
 - 6. Internasal
 - 7. Nasal
 - 8. Premaxillomaxillary and intermaxillopalatine
 - 9. Maxillozygomatic
 - 10. Zygomatic
 - 11. Zygoauromandibular
 - 12. Temporoaural
 - 13. Temporoauromandibular
 - 14. Mandibular
 - 15. Intermandibular
 - C. With synostosis
 - 1. Craniosynostosis
 - 2. Craniofaciosynostosis
 - 3. Faciosynostosis
 - D. With dysostosis and synostosis
 - E. With dyschondrosis
- IV. Craniofacial Dysplasia With Other Origin

subdivided by dysplasias with clefting, dysostosis, and synostosis. For example, the type II oblique cleft of Boo-Chai⁹⁷ is replaced by the lateronasomaxillary cleft under the group of craniofacial dysplasias and holoprosencephaly and by rhinencephalic dysplasia in the group of cerebrofacial dysplasias.

Unfortunately, the terms dysplasia, dysostoses, and synostoses, used in the scheme, are not well defined. The accepted meaning of the term dysplasia now refers to generalized disorders of skeletal involvement, and the term dysostosis refers to involvement of a few regional bones (International Nomenclature of Constitutional Diseases of Bone). The value of replacing the term *mandibulofacial dysostosis* (Treacher Collins syndrome) with *zygomaticotemporoauromandibular dysplasia* is unclear.^{10,101}

Nevertheless, a classification scheme, including both descriptive terminology and insights into the underlying embryologic cause, would be preferable. However, Tessier's simplistic classification, despite its disregard for any embryologic or pathologic sequence, remains the most useful and widely accepted classification scheme today.

TESSIER CLASSIFICATION

In 1974 at the Interdisciplinary Workshop Conference in Chicago on Craniofacial Surgery, Tessier^{98,102} presented a comprehensive classification scheme of craniofacial clefts based on personal experience with 336 patients. Detailed descriptions were then later published in 1976 by Tessier^{98,102} and Kawamoto.⁹

Tessier's system^{9,10,98,102} represents an architectural description of defects rather than offering true descriptions of the structures involved. The defects are numbered for ease of clinical classification rather than along any pathoembryogenic sequence. Although it fails to provide insight into the underlying etiopathology of these defects, the relative simplicity of his classification has made it almost universally accepted for clinical use. The classification has significant value to the practicing reconstructive surgeon, because it is based not only on careful physical examination and radiologic studies, but also correlated with anatomic findings at the time of surgery.

Tessier^{9,10,98,102} noted that the important landmarks through which the fault lines cross are the mouth, nose, orbit, and cranium and found that 15 distinct cleft locations can be differentiated. The clefts are numbered from 0 to 14, in a counterclockwise fashion, circumferentially around the orbit, along constant planes (Figs. 26-13 through 26-16). The orbit was selected as the point

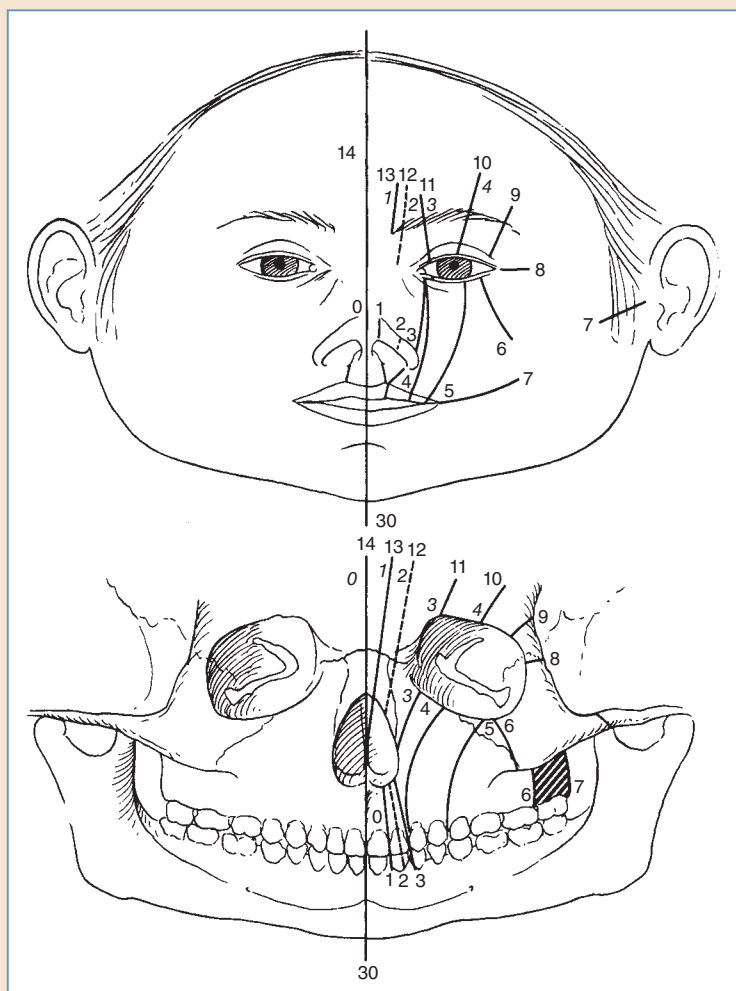


Fig. 26-13 Tessier proposed a comprehensive architectural classification of atypical clefts in 1974. The clefts are centered on the orbit and are numbered from 0 to 14. The soft tissue clefts are correlated with the corresponding skeletal fault lines. (Courtesy of Paul Tessier, MD.)

Fig. 26-14 Detail of the clefts involving the upper lip and nostril and the corresponding anterior dentoalveolar clefts. Clefts 1, 2, and 3 have their common origin at the Cupid's bow peak and correspond to the more prevalently seen cleft lip deformity. Beginning with the number 4 cleft, the ala and piriform aperture are spared. (Courtesy of Paul Tessier, MD.)

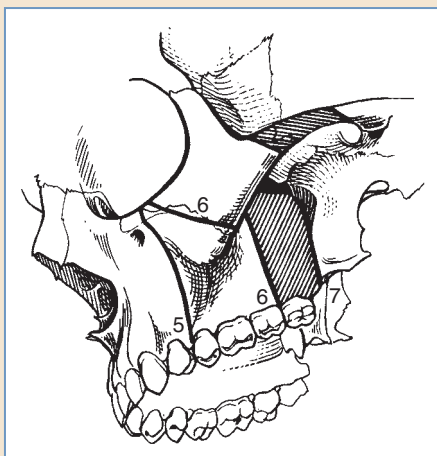
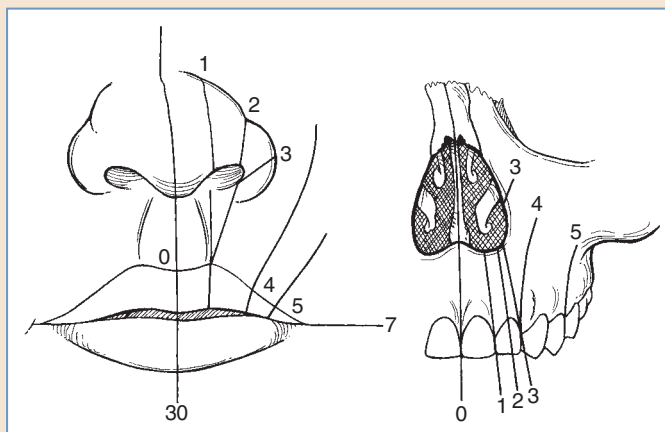


Fig. 26-15 Detail of clefts involving the posterior dentoalveolar region. The number 5 cleft originates in the premolar region. A cleft of the posterior maxillary alveolus is occasionally seen with the number 7 cleft. In contrast, a cleft, per se, is not seen with the number 6 cleft but instead an area of alveolar hypoplasia. (Courtesy of Paul Tessier, MD.)

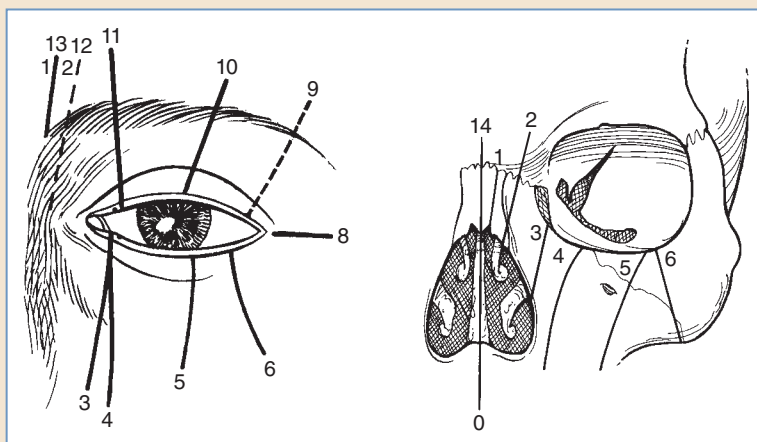


Fig. 26-16 Detail of clefts involving the orbital region. The course of the clefts is shown in relation to the eyelids and eyebrow, along with the corresponding involvement of the underlying orbital skeleton. (Courtesy of Paul Tessier, MD.)

of reference, because it belongs to both the cranium and face. These planes then divide either the cranium or face, depending on the direction the cleft takes from the orbit or palpebral fissure. Southbound clefts pass caudally from the orbit and become purely facial clefts, and northbound clefts passing cephalad from the orbit become cranial clefts.^{9,10,98,102}

Cranial (northbound) and facial (southbound) clefts frequently coexist, and when they do, their axes usually (although not always) follow the same general direction. Such craniofacial clefts are then numbered by their component parts. For example, facial cleft number 0 is seen with its cranial component 14; similarly, combinations of clefts 1 through 13, 2 through 12, 3 through 11, 4 through 10, and 5 through 9 are clinically observed. The combinations of cranial and facial (craniofacial) clefts usually add up to 14. Combinations of purely facial clefts also exist. Treacher Collins syndrome is a combination of clefts 6, 7, and 8.^{9,10,98,102}

Tessier's numeric system is purely a topographic map of the cleft fault lines. It describes neither the specific structures involved nor the degree of involvement of each of the structures. The face can be marred by both a mild crease in the skin or a subtle change in the contour of the hairline to a frank expression of the cleft (Figs. 26-17 and 26-18). Both are described by the same

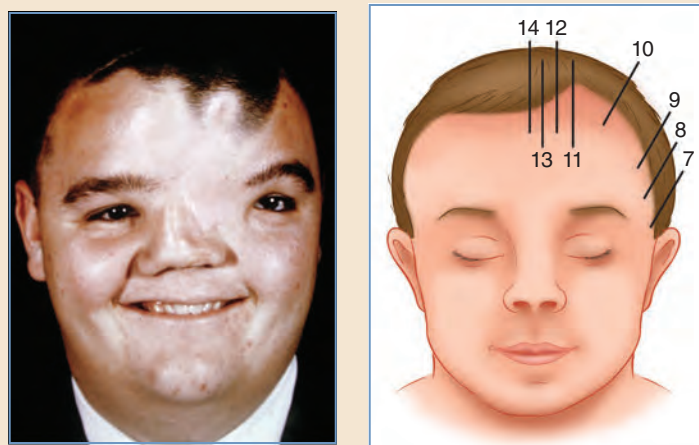


Fig. 26-17 Disruption of hairlines is often seen with the cranial extension of the clefts, and as a subtle finding, they may suggest the course of the cleft. (Courtesy of J.M. Converse, MD.)

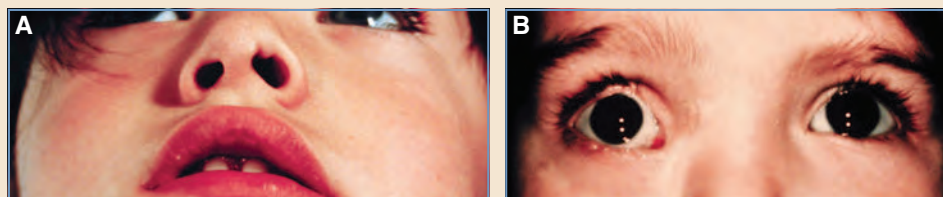


Fig. 26-18 Clefts do not need to be expressed fully and may be marked only by subtle soft tissue disruptions. **A**, An alar notch suggests a number 1 cleft. **B**, Colobomas and dystopia of the medial and central eyelashes and eyebrow suggest a number 10 cleft.

number in the classification. For example, a number 7 cleft describes a situation in which there is only a transverse cleft of the oral commissure, resulting in macrostomia. It also describes a more complete form in which all structures along its path, both soft tissue and skeletal, are involved, resulting in the typical features of hemifacial microsomia.^{9,10}

The value of Tessier's classification scheme is that it directs the clinician to search for the malformations along the entire axis. The structures that may be involved in a particular cleft are then inferred from the path that the cleft takes. For example, without careful examination of the cephalic structures of the common cleft lip, malformations of a more complex cleft can be missed. Both the common cleft lip and the labial cleft of Tessier's facial cleft numbers 1, 2, and 3 begin in the region of the Cupid's bow. The cranial or northbound clefts not previously suspected may be revealed by subtle irregularities of the anterior hairline.^{9,10,103}

In Tessier's original line drawings,⁹⁸ both clefts 2 and 9 were drawn with dotted lines. Both clefts were notably rare. Tessier based the axis of the number 9 cleft on cases reported by Morian⁹³ and Sanvenero-Rosselli,⁹⁴ and Tessier was unsure whether cleft 2 represented a distinct entity. Since the original descriptions, the clefts have been identified in the literature.^{8,104-107}

Description of Facial Clefts

Although the upcoming section describes the individual clefts, there are general rules of thumb that will be helpful. Table 26-4 summarizes the comparative anatomy of the Tessier cleft pattern. Clefts 0, 1, and 2 course medial to the canthus, and as such, do not directly pass through or disrupt the orbit. Clefts 1, 2, and 3 begin at the Cupid's bow, as does the common cleft lip. The distinguishing features are then markedly more cephalic in the nasal region. Cleft 1 has a characteristic notch of the alar dome, and cleft 3 undermines the alar base in its course to involve the orbit. The upper lip cleft of 4 begins lateral to the philtrum, and cleft 5 begins just medial to the commissure. Neither cleft 4 nor 5 directly involves the piriform aperture or the nasal structures. Clefts 3 and 4 enter the palpebral fissure medial to the inferior punctum of the canalicular system, and as such, disrupt the normal anatomy of the medial canthal region. Cleft 5 enters lateral to the punctum, and the medial canthus is not directly involved. Cleft 4 courses medial to the inferior orbital foramen, and cleft 5 courses lateral to the foramen. Cleft 6 has no labial component and is purely an orbital cleft, whereas cleft 7 has no orbital component and labially is a transverse cleft, originating from the oral commissure. Cleft 8 originates as a transverse cleft from the lateral commissure of the palpebral fissure. Beginning with the number 9 cleft, the cleft becomes cranial or northbound. Clefts 9, 10, and 11 disrupt the upper eyelid and eyebrow. Clefts 10 and 11 can result in orbital dystopia because of direct involvement of the orbit. Clefts 12, 13, and 14 course medial to the orbit and result in hypertelorism.

From the midline to the inferior orbital foramen, the clefts of the soft tissue are more frequent and more disruptive than those of the underlying skeleton. In contrast, from the foramen laterally, the defects in the bony structures are typically more involved than those of the soft tissue. Two notable exceptions are the more frequent soft tissue auricular involvement of the number 7 cleft and the more significant skeletal disruption of the number 3 cleft that can occur.

The most common cleft types seen are the median clefts 0 and 14, although the transverse cleft, number 7, would be expected to be significantly more common with a stricter definition. The least frequently encountered are oblique cleft numbers 4, 5, 8, and 9 of the atypical clefts. In contrast, paramedian clefts are encountered more often than oblique clefts.

Table 26-4 Comparative Anatomy of Atypical Craniofacial Clefts

Cleft Type	Lip Alveolus	Nasal Region Piriform Aperture	Cheek Maxilla/ Zygoma	Orbital Level	Supraorbital/Cranium	Cleft Type
0	True median cleft Between central incisors	Bifid nose Septum thick, duplicated	— —	— Nasal bones hyperplastic Ethmoid labyrinth Hypertelorism	Flattened frontal region or median encephalocele Bifid cranium Crista galli widened, duplicated Distance between olfactory grooves increased Ethmoid labyrinth	14
1	Cupid's bow Between central/lateral incisors	Alar dome notch Piriform aperture lateral to anterior nasal spine	— —	— Nasal bone Ethmoid labyrinth Hypertelorism	Dystopia of the medial edge of brow Paramedian encephalocele Widened olfactory groove/cribriform plate Ethmoid labyrinth	13
2	Cupid's bow Region of lateral incisor	Middle third alar rim Lateral piriform aperture	— —	— Nasal bone/frontal process of maxilla Ethmoid labyrinth Hypertelorism	Disrupts medial third of brow Upper eyelid uninvolved Lateral to the cribriform plate Ethmoid labyrinth	12
3	Cupid's bow Between lateral incisor/cuspid	Alar base Junction of lateral nasal wall/floor	Lateral nasal-cheek junction Loss of normal bony septation between the nasal/maxillary cavity	Medial canthus disrupted Lower eyelid defect medial to the punctum Lacrimal system disrupted 1. Frontal process of the maxilla Ethmoid labyrinth Hypertelorism 2. Lateral to the ethmoid bone	Medial third of upper eyelid Medial third of the eyebrow 1. Lateral to the cribriform plate Ethmoid labyrinth or 2. Medial third of the supraorbital rim	11
4	Midway between philtrum and commissure Between lateral incisor/cuspid	— —	Medial cheek Anterior maxillary wall medial to the infraorbital foramen	Lower eyelid medial to the punctum Lacrimal system except for inferior canaliculus is spared Inferomedial orbital rim and floor	Middle third of upper eyelid Middle third of eyebrow Middle third of the supraorbital rim	10
5	Medial to the commissure Distal to the cuspid in the premolar region	— —	Lateral cheek Anterior maxillary wall lateral to the infraorbital foramen	Medial and lateral third of the lower eyelid Middle third of the orbital rim and floor	Lateral third of upper eyelid Lateral third of eyebrow Superolateral orbital rim	9
7	Oral commissure Incomplete cleft molar region	— —	Lateral cheek to the ear Zygomatic arch Mandible	Medial and lateral third of the eyelid Antimongoloid slant palpebral fissure Zygomaticomaxillary suture	Lateral canthus Frontozygomatic suture	8
Cleft Type	7			6	8	Cleft Type

NOTE: Clefts 0-14 include "median hypoplasia" (holoprosencephaly), not included in the above table.

Based on Tessier's classification, the most reliable figures in the distribution of the types of clefts come from the large data collected by Ortiz-Monasterio and Taylor⁸ (Table 26-5). In their series, cleft 0 accounted for 17% of the cases, cleft 7 for 15% of the cases, and cleft 14 for 12% of the cases. The least common atypical clefts reported were the number 5 and 9 clefts (<1%). Moreover, craniofacial clefts do not necessarily occur as isolated clefts and are frequently combined with other clefts (Table 26-6).

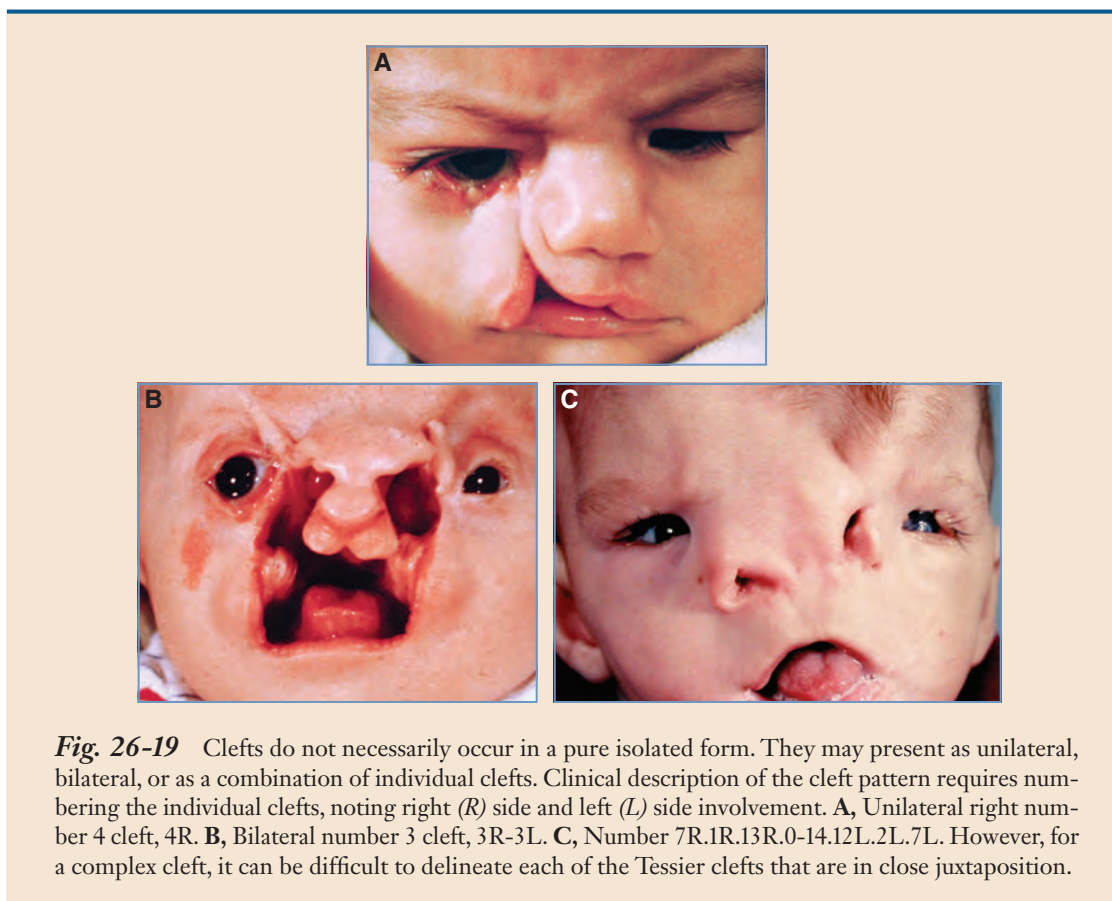
Table 26-5 Atypical Craniofacial Clefts

Tessier Number	Presenting as Single Cleft	Presenting as Multiple Clefts	Total (N = 998)	Percentage of Total
0	40	132	172	17.2
1	5	81	86	8.6
2	14	47	61	6.1
3	20	48	68	6.8
4	12	26	38	3.8
5	3	5	8	0.8
6	2	57	59	5.9
7	58	89	147	14.7
8	6	58	64	6.4
9	—	5	5	0.5
10	3	17	20	2.0
11	2	22	24	2.4
12	1	37	38	3.8
13	2	81	83	8.3
14	3	121	124	12.4

Series of 998 clefts in 495 patients between 1965 and 2006.

Table 26-6 Incidence of Atypical Multiple Cleft Pattern

Tessier Number	Number of Multiple Presentations	Percentage of Multiple Presentations	Percentage of Total Clefts
0-14	115	35.7	11.5
0-13	3	0.9	0.3
1-13	71	22.0	7.1
2-12	31	9.6	3.1
3-11	10	3.1	1.0
3-12	1	0.3	0.1
6-7-8	52	16.1	5.2
Various	39	12	3.9



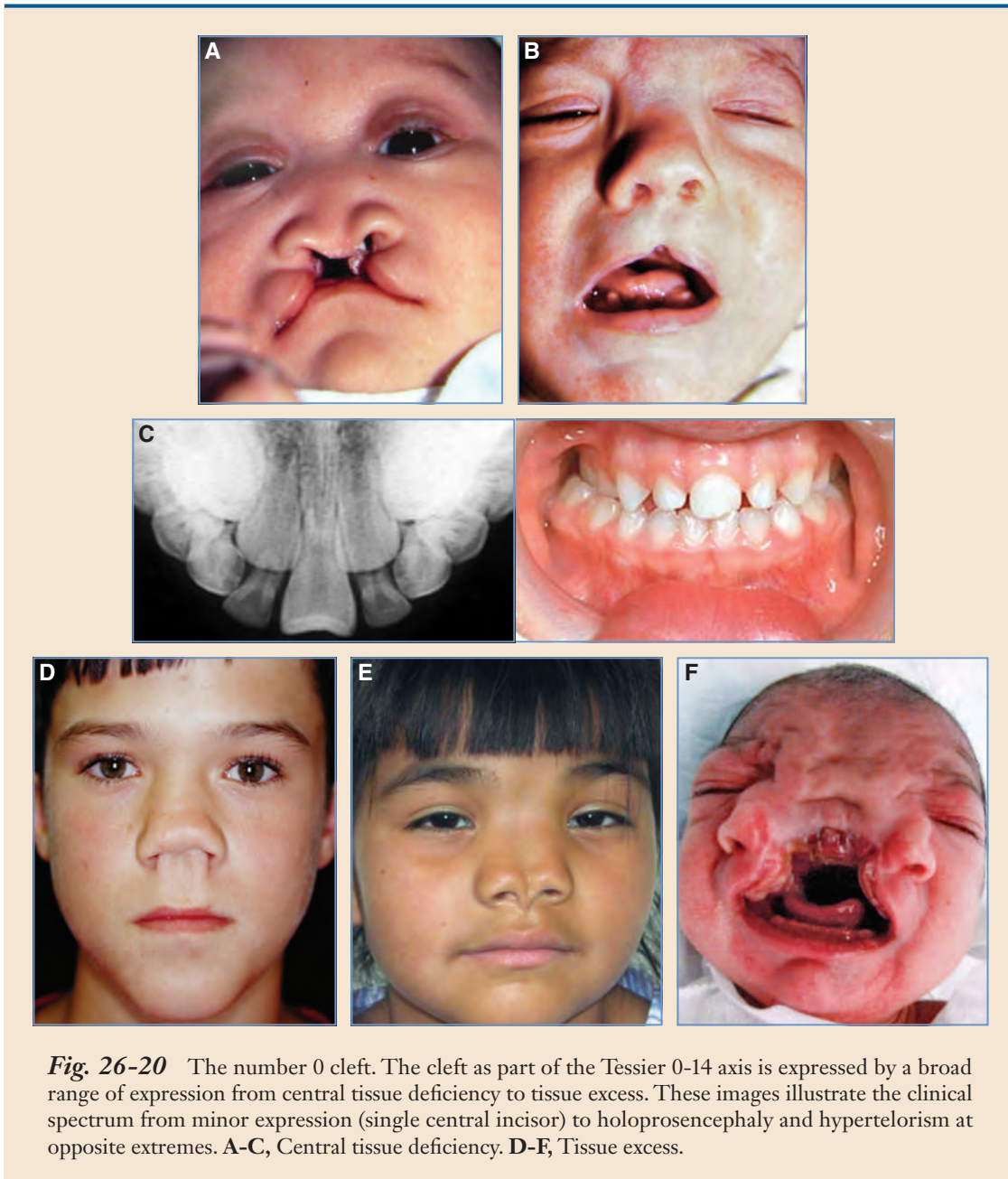
The clinical presentation of a cleft pattern may be considered as a combination of separately numbered elements of the Tessier system (Fig. 26-19). The most common combination was 0-14 (36%), followed by 1-13 (22%) and the 6-7-8 combination (16%).

Unilateral and bilateral clefts can be found in varying combinations (see Fig. 26-19). In their series of 495 patients, the atypical cleft was unilateral in 164 patients (33%) and bilateral in the remaining 331 (67%) of patients. When clefts are bilateral they are frequently symmetrical with respect to their axis, but not necessarily in the severity of their involvement. Nearly one half of the cases of clefts 1 and 2 present as bilateral clefts. In contrast, the more oblique clefts 3, 4, and 5 present less frequently, between 20% and 30%, as bilateral clefts.^{8,10}

Number 0 Cleft

The number 0 cleft is in the midline of the face (Fig. 26-20). Along with its cranial extension, the number 14 cleft, it is a median craniofacial dysraphia and includes the median cleft face dysmorphism (hyperplasia) and holoprosencephaly (hypoplasia). The 0-14 axis can be further classified along the lines previously discussed.

The cleft of the upper lip varies from a minor vermillion notch to a true median cleft. The frenulum is often duplicated, and there is a diastema between the maxillary central incisors. The nose is frequently bifid, and the columella and dorsum of the nose are wide and often associated with a median furrow. Its skeletal course follows a midline path. The bony cleft begins in the alveolus between the central incisors. The dentoalveolar arch is angulated upward into the cleft.



Posteriorly, the cleft may continue in the midline of the premaxilla through the secondary palate. Superiorly, the anterior nasal spine and septum may be either thickened or duplicated. The lower and upper lateral cartilages of the nose are displaced laterally and may be hypoplastic or distorted. The nasal bones are wide and flattened. The anterior ethmoid cells are increased in number and enlarged (posterior ethmoid and sphenoid are typically uninvolved), resulting in hypertelorism.*

*References 9, 10, 91, 98, 102, 104, 108-113.

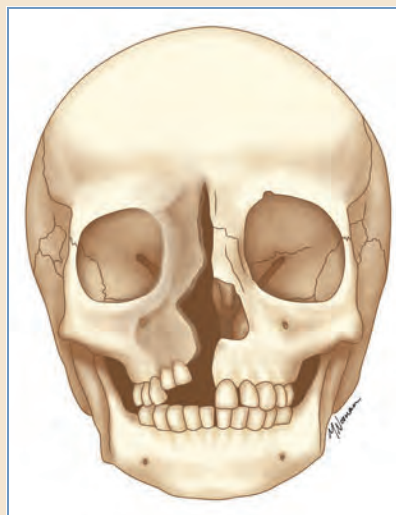


Fig. 26-21 The number 1 cleft. The characteristic finding of the cleft is notching of the ala dome. The cleft begins in the region of the lateral and central incisors and courses through the nasal floor and superiorly through the nasal bones. The cleft continues cranially as a number 13 cleft.

At the other end of the spectrum, the number 0 cleft may have the hypoplastic features of holoprosencephaly. A false median cleft of the lip is seen with partial or total absence of the probolabium. The nose may be rudimentary, absent (arhinia), or merely a proboscis. The midline bony defect results in either partial or complete absence of the premaxilla. A cleft of the secondary palate is often seen. The septum, if present, is vestigial. The osteocartilaginous portion of the nasal vault may be hypoplastic or entirely absent. Superiorly, the bony defect continues as a number 14 cleft into the ethmoid, resulting in hypertelorism.*

Number 1 Cleft

The Tessier number 1 cleft begins in the region of the Cupid's bow, continues northbound to involve the alar dome of the nostril, and extends onto the nasal dorsum. Although telecanthus has been described, the fault passes well medial to the canthal structures, which are normal (Fig. 26-21).

The skeletal course of the cleft passes through the alveolus between the central and lateral incisors, through the base of the piriform aperture, just lateral to the anterior nasal spine without involving the septum directly. The cleft of the maxilla may extend posteriorly to form a complete cleft of the hard and soft palates. Superiorly, the fault continues through the nasal bone medial to its junction with the frontal process of the maxilla. Hypertelorism results as the fault courses parasagittally toward the glabella-forehead region to involve the ethmoidal labyrinth. It continues as a Tessier number 13 cleft with involvement of the cranium. The combination 1-13 cleft is a paramedian craniofacial cleft.^{9,10,98,102,104}

*References 9, 10, 82, 98, 102, 104, 114-116.

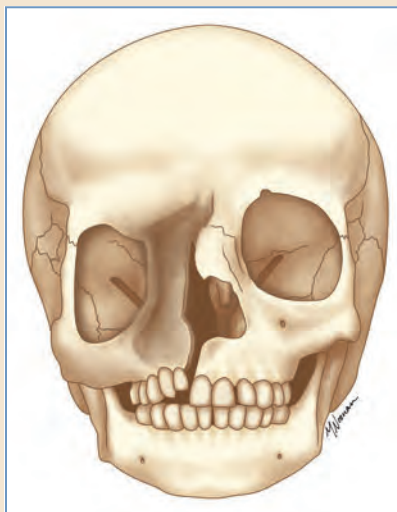


Fig. 26-22 The number 2 cleft. A characteristic of the cleft is that the alar rim appears hypoplastic and is drawn upward. A true notch, per se, of the alar rim is not seen. The cleft involves the frontal process of the maxilla and continues superiorly as a number 12 cleft.

Number 2 Cleft

The Tessier number 2 cleft begins in the region of the Cupid's bow and continues superiorly through the middle third of the nostril rim (Fig. 26-22). Unlike the notched dome of the number 1 cleft, the alar involvement has a hypoplastic appearance, resulting in more of a flattened appearance of the lateral aspect of the nose. Superiorly, the cleft courses as a soft tissue groove to an asymmetrically broadened nasal root with displacement of the inner canthus. The lacrimal system, palpebral fissure, and eyebrows remain uninvolved.

In the alveolus the fault begins in the lateral incisor region to involve the lateral portion of the base of the piriform aperture. Unlike the number 3 cleft, the lateral nasal wall is spared, and the maxillary sinus is not in continuity. Superiorly, the cleft courses obliquely through the junction of the nasal-frontal process of the maxilla without involving the orbital rim. The medial canthus and palpebral fissure are uninvolved. As the cleft comes through the lateral ethmoidal labyrinth, hypertelorism results. Its cranial extension is the number 12 cleft.^{9,10,92,102,104}

Number 3 Cleft

Similar to the number 2 facial cleft, the Tessier number 3 cleft begins as a common cleft of the lip. However, as it continues cephalad, the cleft passes through the nasal alar-cheek junction and continues along the lateral nasal margin to involve the medial aspect of the lower eyelid, just medial to the inferior nasolacrimal punctum (Fig. 26-23). As such, the lower canalicular system is malformed and beyond repair, and the nasolacrimal drainage is disrupted and prone to recurrent episodes of infection. Unlike the number 2 cleft, the medial canthus is involved; it is displaced inferiorly and the canthal ligament is hypoplastic. As a result of the cleft, there is significant soft tissue deficiency in the vertical dimension between the alar base and lower eyelid. The ocular globe may be normal but dystopic or microphthalmic. As the cleft courses superiorly, the medial third of the upper eyelid and brow may be involved as a number 11 cleft.

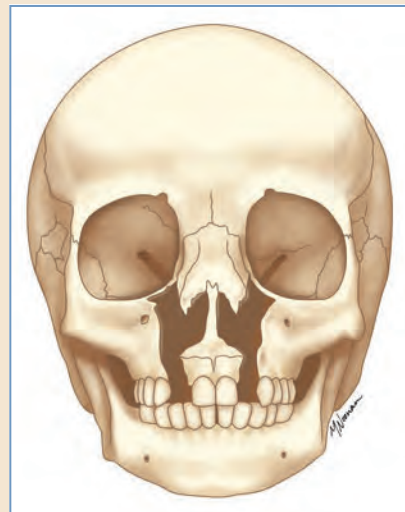
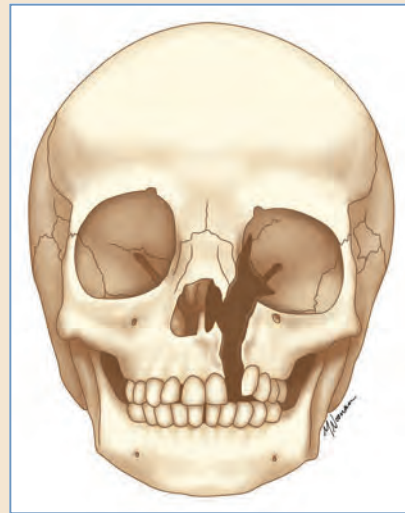


Fig. 26-23 The number 3 cleft. The cleft undermines the alar base and continues superiorly, medial to the inferior punctum, and interrupts the nasolacrimal system to involve the medial canthal structures. The cleft disrupts the lateral nasal wall and orbital floor. As a result, the orbital, maxillary, and nasal cavities are in continuity. The cleft continues cranially as a number 11 cleft.

As in the typical alveolar cleft, the skeletal cleft passes between the lateral incisor and canine, but proceeds to disrupt the lateral border of the piriform aperture to involve the medial wall of the maxillary sinus. In contrast to the number 2 cleft, the number 3 cleft continues superiorly to involve the frontal process of the maxilla to the lacrimal groove, involving the medial orbital floor and rim. As such the skeletal disruption can be extensive, with the normally separated cavities of the mouth, nose, maxillary sinus, and orbit in continuity as a result of the cleft. Thus the maxilla is significantly hypoplastic in all three dimensions.*

*References 4, 9, 10, 47, 93, 97, 98, 102, 104, 107, 117-122.

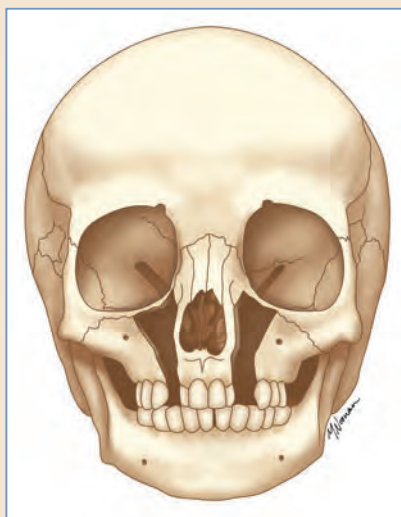
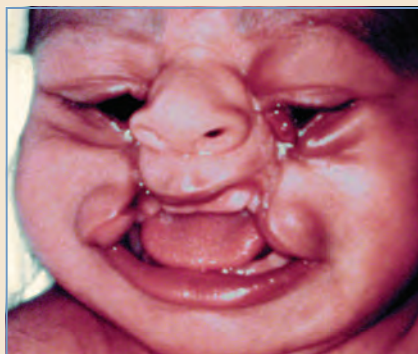
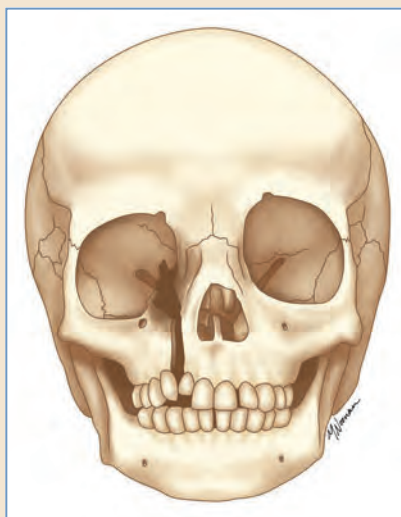


Fig. 26-24 The number 4 cleft. The cleft begins lateral to the Cupid's bow. The ala is undisturbed, although it is often secondarily drawn upward. The cleft continues between the piriform aperture and infraorbital foramen to involve the medial orbital floor. The lateral nasal wall is spared, and only the orbital and maxillary sinus are confluent.

Number 4 Cleft

The number 4 cleft begins midway between the philtral column and the commissure, continues lateral to the nasal ala onto the cheek, and ends in the lower eyelid medial to the inferior punctum (Fig. 26-24). Although the inferior canaliculus is involved, the lacrimal sac and nasolacrimal canal are usually undisturbed. The medial canthal tendon is relatively unaffected, because the path of the fault lies lateral to it. As with the number 3 cleft, there is a severe soft tissue deficiency with the margins of the cleft lip, and the normal ala is drawn into the coloboma of the lower eyelid defect. Similarly, orbital dystopia, which occurs as a result of skeletal involvement and microphthalmia, has been described. Superiorly, the cleft continues as a number 10 cleft to involve the middle third of the upper eyelid and eyebrow.

The bony discontinuity cleft begins between the lateral incisor and canine, courses lateral to the piriform aperture through the anterior maxillary wall, and medial to the inferior orbital

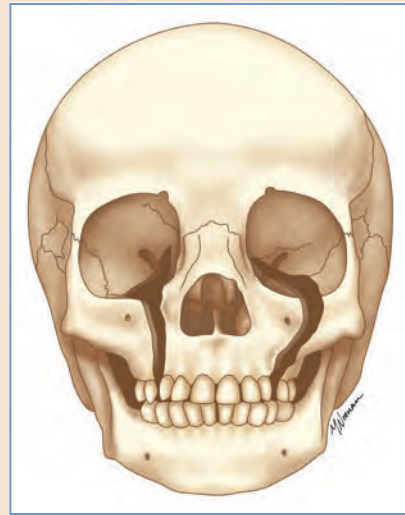


Fig. 26-25 The number 5 cleft. In contrast to the number 4 cleft, the number 5 cleft begins near the oral commissure and courses toward the lateral orbital region. On the facial skeleton, the cleft lies lateral to the infraorbital foramen.

foramen, to end at the medial orbital rim and floor. Unlike the number 3 cleft, the piriform aperture and normal bony septation between the maxillary sinus and nasal cavity are spared, and in its complete form, only the oral cavity, maxillary sinus, and orbit are in continuity.*

Number 5 Cleft

The number 5 cleft begins just medial to the oral commissure and follows a more oblique path than the number 4 cleft onto the lateral aspect of the cheek to terminate within the region of the middle and lateral thirds of the lower eyelid (Fig. 26-25). As a result, the vertical soft tissue deficiency of the midface occurs more laterally between the lateral lip element and lower eyelid cleft. Vertical orbital dystopia and microphthalmia are thought to occur. Superiorly, the cleft continues as the number 9 cleft to involve the lateral third of the upper eyelid and brow. Its osseous path begins in the alveolus distal to the canine in the premolar region and continues superiorly across the anterior maxillary wall lateral to the inferior orbital foramen to enter the orbital rim and floor. As with the number 4 cleft, the oral cavity, maxillary sinus, and orbit may be in continuity.†

Number 6 Cleft

The soft tissue cleft begins at the junction of the middle and lateral third of the lower eyelid and continues inferiorly toward the angle of the mandible, as a crease, to give the appearance of an antimongoloid slant (Fig. 26-26). The external ear is not typically involved, although a hearing deficit is frequently described. Its osseous component is characterized by a hypoplastic zygoma, with the cleft running through the zygomaticomaxillary suture. At the level of the orbital rim and floor, the cleft opens into the inferior orbital fissure. Although a true cleft of the alveolus has not been described, the posterolateral aspect of the maxilla is hypoplastic in the molar region.‡

*References 9, 10, 47, 97, 98, 102, 104, 107, 120-126.

†References 7a, 9, 10, 93, 97, 98, 102, 104, 107, 127, 128.

‡References 9, 10, 98, 102, 104, 128-130.

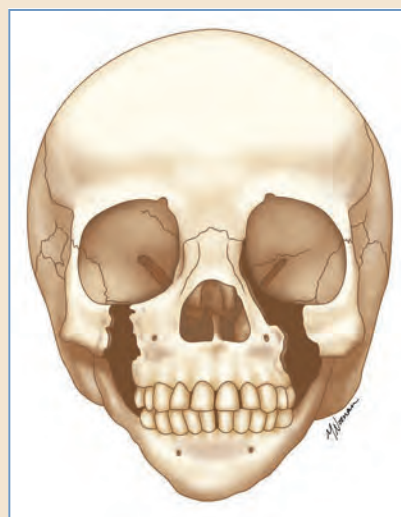


Fig. 26-26 The number 6 cleft. The cleft is recognized as an incomplete form of Treacher Collins syndrome with a coloboma of the lateral lower eyelid and malar hypoplasia. The zygoma is present but hypoplastic, with the cleft in the region of the zygomaticomaxillary suture.

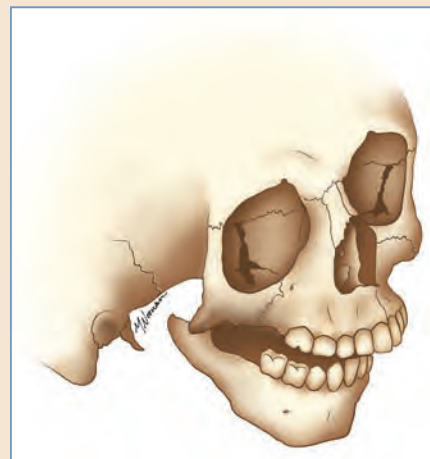
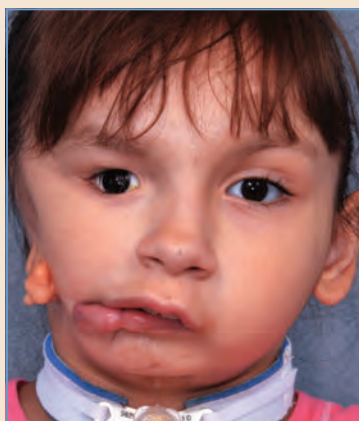
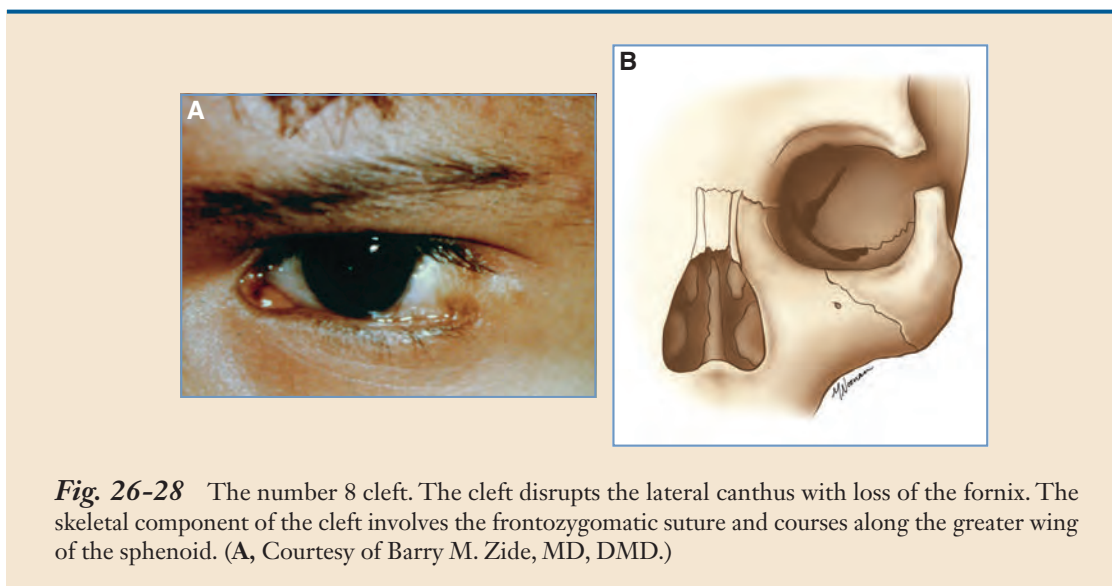


Fig. 26-27 The number 7 cleft. More commonly known as *craniofacial microsomia*, the cleft has a broad spectrum of presentation, with varying degrees of soft tissue and skeletal involvement. The soft tissue component ranges from macrostomia to varying degrees of microtia. The skeletal component of the cleft involves the mandible and zygomatic arch to varying degrees. The maxilla, zygomatic body, orbit, and cranium may be involved. The skeletal component of the number 7 cleft has been further subdivided by classification schemes described by Pruzansky, Munro and Lauritzen, and Vento et al.

Number 7 Cleft

The fault line of the number 7 cleft extends along the transverse crease between the oral commissure and auricular tragus¹³¹⁻¹³³ (Fig. 26-27). The soft tissue deficiency varies from an isolated occurrence of macrostomia or preauricular tags alone, to more severe soft tissue involvement of all intervening structures. The ipsilateral tongue and soft palate may be involved. The parotid gland and duct may be hypoplastic or absent. Occasionally, fifth and seventh cranial nerve palsies can occur. The muscles of mastication—the temporalis, masseter, and pterygoid—may be under-



developed and confluent. The auricular deformities involve not only the external auricle but also the auditory canal and middle ear ossicles.

The skeletal cleft courses between the zygomatic and temporal bones. The zygomatic arch is disrupted, and along with it, the mandibular coronoid process, condyle, and ramus are hypoplastic to varying degrees. The condyle may be distorted or absent altogether. The ramus may be short with a loss of the gonial angle. With the vertical shortening of the mandibular ramal height and posterolaterally the maxilla, the occlusal plane is canted cephalad on the affected side. In the maxillary molar region, the fissure continues between the maxillary tuberosity and pterygoid plate.^{9,10,98,102,104}

Tessier's cleft number 7 is more commonly recognized in the literature as hemifacial microsomia, mandibulofacial dysostosis, and first and second branchial arch syndrome.^{13,129} Goldenhar syndrome is closely associated and has the additional features of epibulbar dermoids and vertebral anomalies.¹³⁰ When bilateral, the number 7 cleft is also found in Treacher Collins syndrome or mandibulofacial dysostosis. The various classification schemes by Pruzansky,¹³¹ Munro and Lauritzen,¹³² Vento et al,¹³³ and others for hemifacial microsomia can be included within the context of the number 7 cleft to further order the degree of involvement.

Number 8 Cleft

The number 8 cleft begins at the lateral canthus and continues posteriorly into the temporal region. The soft tissue defect can be merely a depression at the level of the lateral canthus or a true cleft of the lateral commissure. The defect is characterized by a strip of intervening skin between the upper and lower eyelids, obliterating the depth of the conjunctival fornix (Fig. 26-28). The cleft prevents continuity of the orbicularis oculi muscle and fixation of the lateral canthal tendon. Its osseous component begins in the region of the frontozygomatic suture and can involve the greater wing of the sphenoid.^{9,10,100,104,106}

Facial Clefts 6, 7, and 8 Combination

Tessier^{9,10,98,102} recognized the bilateral combination of clefts 6, 7, and 8 as characterizing the physical findings of patients previously described in the literature with Treacher Collins syndrome or mandibulofacial dysostosis (Fig. 26-29). The course of clefts 6, 7, and 8 isolates the zygoma, and its absence is the hallmark of the complete form of the syndrome. Cleft 6 accounts

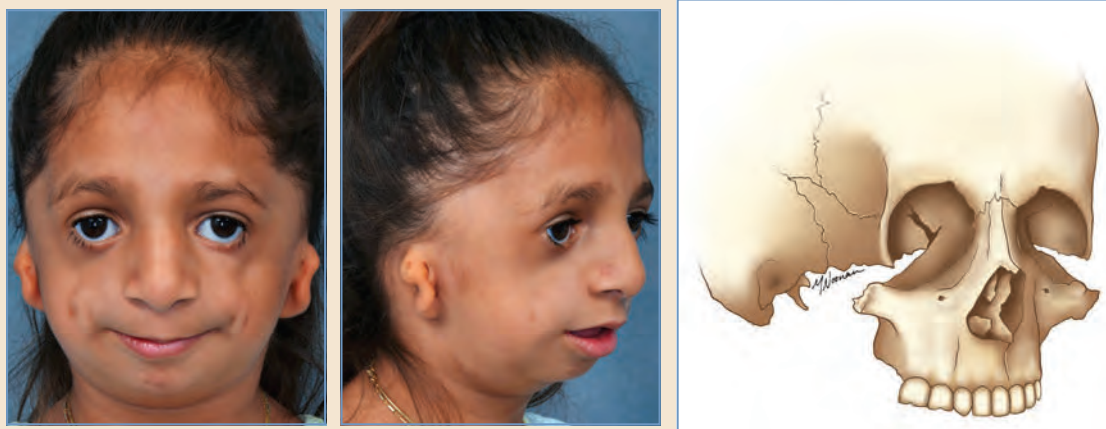


Fig. 26-29 Numbers 6, 7, and 8 cleft combination. The cleft results in an absence of the zygoma, with the greater wing of the sphenoid forming the lateral orbital rim. In the bilateral form, the skeletal deformity is characteristic of the Treacher Collins syndrome malformation: malar hypoplasia, downward slanting (antimongoloid) palpebral fissures and lower eyelid colobomas.

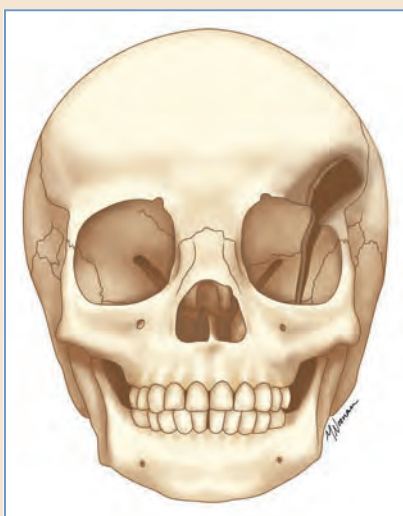


Fig. 26-30 The number 9 cleft. The cleft begins in the superolateral portion of the orbital roof through the greater wing of the sphenoid and courses more obliquely to involve the frontal, temporal, and parietal bones.

for the eyelid coloboma and bony hypoplasia at the zygomaticomaxillary suture. The number 7 cleft contributes to the auricular malformation and macrostomia defects, fusion of the temporalis and masseter muscles, and the osseous zygomaticotemporal and mandibular deformities. Cleft 8 completes the syndrome with the zygomaticofrontal (lateral orbital wall) deformity and antimongoloid slant with loss of lateral canthal fixation. Incomplete forms of the syndrome can then be described by various combinations of the number 6, 7, and 8 clefts.^{9,10,98,102}

Number 9 Cleft

Beginning with the number 9 cleft, the clefts become northbound and involve the superior orbital and frontal cranial region. The osseous fault of the number 9 cleft begins in the superolateral portion of the orbital roof through the greater wing of the sphenoid and courses more obliquely to involve the frontal, temporal, and parietal bones (Fig. 26-30). Likewise, the path of the soft

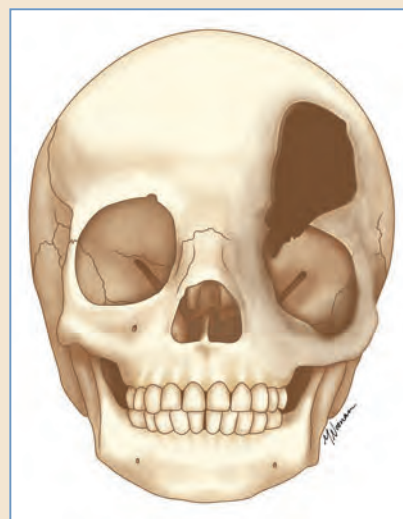


Fig. 26-31 The number 10 cleft. The cleft courses through the central upper eyelid, with colobomas of the eyelash and brow. The central orbital roof and frontal bone are disrupted, and the cranial defect is often occupied by a frontoorbital encephalocele, resulting in an inferolateral orbital dystopia. Its southbound extension is described by the number 4 cleft.

tissue defect courses through the lateral third of the upper eyelid, as does the eyebrow, and into the temporal hairline. The cleft is regarded as the cranial extension of the number 5 cleft.*

Number 10 Cleft

The number 10 cleft involves the middle third of the supraorbital rim, involving the frontal bone and orbital roof lateral to the supraorbital foramen (Fig. 26-31). An encephalocele is often associated with the orbital cleft, resulting in secondary orbital dystopia.

Likewise, soft tissue involvement results in a coloboma of the middle third of the upper eyelid and brow. The extent of lid involvement is variable. Ablepharia has been described, as well as an attempted duplication of the medial third of the lid margin presenting as a transverse split. Ocular anomalies, including colobomas of the iris, may also be associated with this cleft. The frontotemporal hairline may be disrupted in the course of the cleft.^{9,10,98,102,104}

Number 11 Cleft

The number 11 cleft is the cranial extension of the number 3 cleft. As the bony discontinuity continues cephalad, the osseous path can either take a more medial route through the ethmoidal labyrinth or a course lateral to the ethmoid bone to disrupt the medial third of the orbital rim (Fig. 26-32). The medial course results in a unilateral hypertelorism, and the more lateral course results in a coloboma of the medial third of the upper eyelid and proceeds to involve the corresponding medial third of the eyebrow and the frontal hairline.^{9,10,98,102,104}

Number 12 Cleft

As the number 2 cleft courses superiorly to involve the cranium, it continues as a Tessier number 12 cleft. The cleft passes between the junction of the nasal-frontal process of the maxilla and nasal bone to involve the ethmoidal labyrinth, resulting in hypertelorism (Fig. 26-33).

*References 9, 10, 92, 94, 98, 102, 104-107.

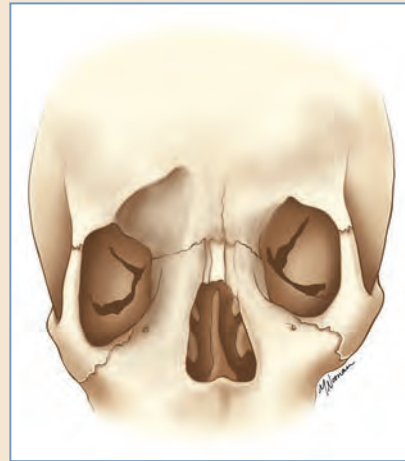


Fig. 26-32 The number 11 cleft. The cleft disrupts the medial third of the upper eyelid and brow. Its osseous path results in either clefting of the medial supraorbital rim or hypertelorism, with involvement of the ethmoidal labyrinth. Its southbound extension is described by the number 3 cleft.

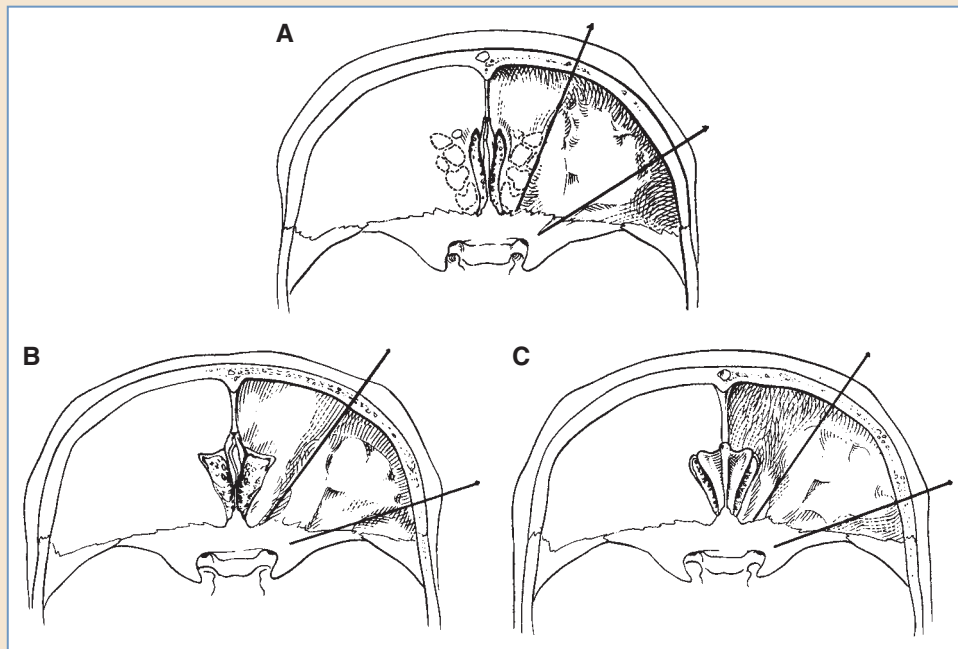


Fig. 26-33 Intracranial view of hypertelorism. **A**, Widening of the ethmoidal labyrinth is seen with the number 12 cleft. **B**, Widening of the olfactory groove and cribriform plate occurs with the number 13 cleft. **C**, The width of the crista galli is greater in the number 14 cleft. (Courtesy of Paul Tessier, MD.)



Fig. 26-34 The number 13 cleft. The cleft courses superiorly medial to the eyebrow as the cranial extension of the number 1 cleft. Hypertelorism is a common feature, with widening of the olfactory groove and cribriform plate.

Unlike the Tessier number 13 cleft, the skeletal fault is lateral to the olfactory groove, and the cribriform plate is of normal width. Although the medial canthus and upper lid are uninvolved, the cleft may disrupt the medialmost portion of the eyebrow.^{9,10,96,102,104}

Number 13 Cleft

The number 13 cleft is the northbound or cranial extension of the paramedian number 1 cleft. The fault lies medial to the junction of the nasal bone and frontal process of the maxilla, to involve the ethmoidal labyrinth and olfactory groove, resulting in widening of the cribriform plate and hypertelorism (Fig. 26-34; see Fig. 26-33). The characteristic finding is a widening of the olfactory groove.

The medial portion of the eyebrow can be dystopic without an obvious crease, and often there is an omega-shaped distortion of the hairline as the underlying skeletal cleft enters the cranium.^{9,10,98,102,104}

Number 14 Cleft

The number 14 cleft returns to the midline of the cranium. As with its facial counterpart, the number 0 cleft, it is part of the median craniofacial dysraphia and includes the median cleft face syndrome (hyperplasia) and holoprosencephaly (hypoplasia) (Fig. 26-35; see Fig. 26-33).

The latter situation is characterized by hypotelorism and the previously discussed features of holoprosencephaly. The central midline cranial base structures may be hypoplastic to varying degrees, and the orbits allowed to converge to the midline. The cranium is microcephalic, and the forebrain abnormality is proportional to the degree of facial disorganization.

Hypertelorism, which is at the other end of the spectrum and more commonly seen, has an increased width in the interorbital distance. The cranium may be bifid. The crista galli may be

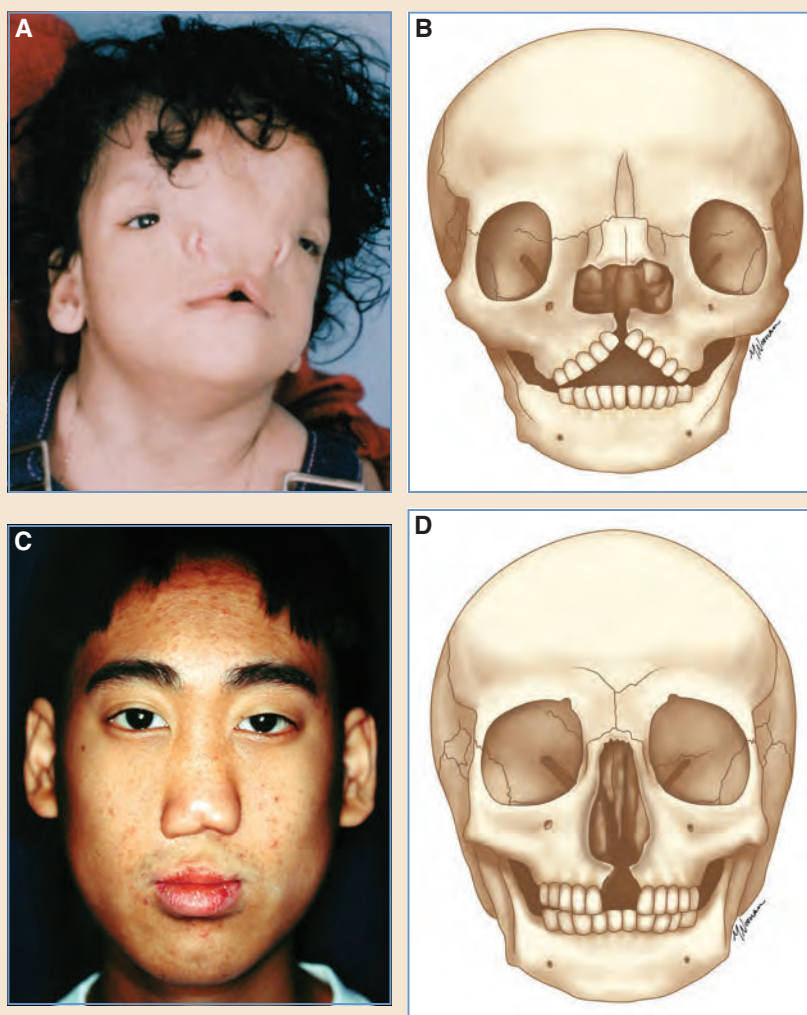


Fig. 26-35 The number 14 cleft. **A** and **B**, The number 14 cleft is the cranial extension of the number 0 cleft, and as such, may present with tissue excess (hypertelorism). **C** and **D**, Alternatively, it may present with tissue deficiency (hypotelorism).

widened, duplicated, or absent. The distance between the olfactory grooves is increased, but the olfactory groove itself is uninvolved and of normal width. Unlike the number 13 cleft, the cribriform plate is of normal width. The bony defect may give rise to a frontal, frontonasal, or frontoethmoidal encephalocele. As a result, the cribriform plate may be inferiorly displaced as much as 20 mm below the level of the orbital roof, when it is normally only 5 to 10 mm. The frontal bone may be flattened or with a central defect when an encephalocele is present. The radix is broad, and the nasal bone is splayed out as the defect courses southbound in continuum with the number 0 cleft.*

*References 9, 10, 98, 102, 104, 105.

Table 26-7 Tessier 0-14 Classification of Median Craniofacial Dysplasia

Tessier 0-14 Axis	Description
I. Median Craniofacial Hypoplasia	Tissue deficiency
A. Holoprocerephaly spectrum (alobar brain)	Single holistic brain with midline facial hypoplasia or agenesis
1. Cyclopia	Single eye in a single orbit, arhinia with proboscis, often located above the single orbit and microcephaly
2. Ethmocephaly	Severe hypotelorism but separate orbits, arhinia with proboscis located between the orbits
3. Cebocephaly	Severe hypotelorism, proboscis-like rudimentary nose
4. Primary palatal agenesis	Hypoplastic or agenesis of the premaxilla
B. Median cerebrofacial hypoplasia (lobar brain)	Separate lobes to the brain but with midline cerebral malformation; midline facial hypoplasia
C. Median facial hypoplasia	Midline facial hypoplasia without gross cerebral involvement
D. Microforms of median facial hypoplasia	Binder anomaly Central single maxillary incisor Absent upper lip frenulum
II. Median Craniofacial Dysraphia	Normal tissue volume
A. True median cleft	Isolated cleft of the upper lip or abnormal split between the median globular process; can be an incomplete or complete form
B. Anterior encephalocele	Cystic malformation in which the CNS is abnormally displaced or herniated through a defect in the cranium
III. Median Craniofacial Hyperplasia	Tissue excess Tissue excess or duplication All forms of excess tissue starting from thickened or duplicated septum to severe forms of frontonasal dysplasia

Numbers 0-14 Craniofacial Cleft Axis

The median craniofacial dysplasias reviewed in the previous section have been subclassified previously by DeMyer et al⁸⁰ in Tables 26-2 and 26-3. The recent reclassification (Table 26-7) by Kawamoto⁹ may provide a more orderly organization of this grouping of patients along the Tessier 0-14 axis from tissue deficiency to tissue excess.¹³⁴

Number 30 Cleft

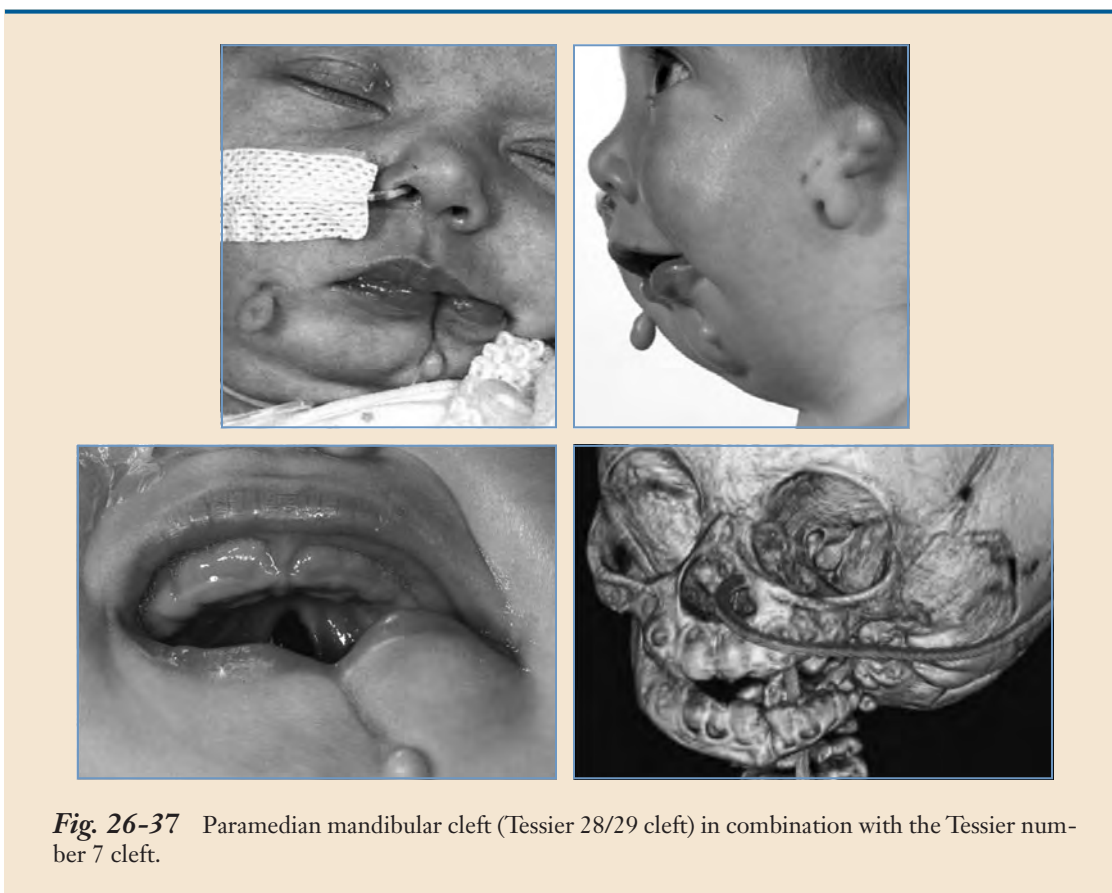
Tessier tentatively labeled the median cleft of the mandibular process as the number 30 cleft to leave the classification scheme open to also include additional paramedian clefts. It is the caudal extension of the number 0-14 cleft. The central defect in the lower lip may range from a minor



Fig. 26-36 The number 30 cleft. Varying degrees of involvement of the median mandibular symphyseal cleft have been reported in the literature, ranging from the commonly seen cleft chin to involvement of the lower lip, alveolus, tongue, and cervical regions.

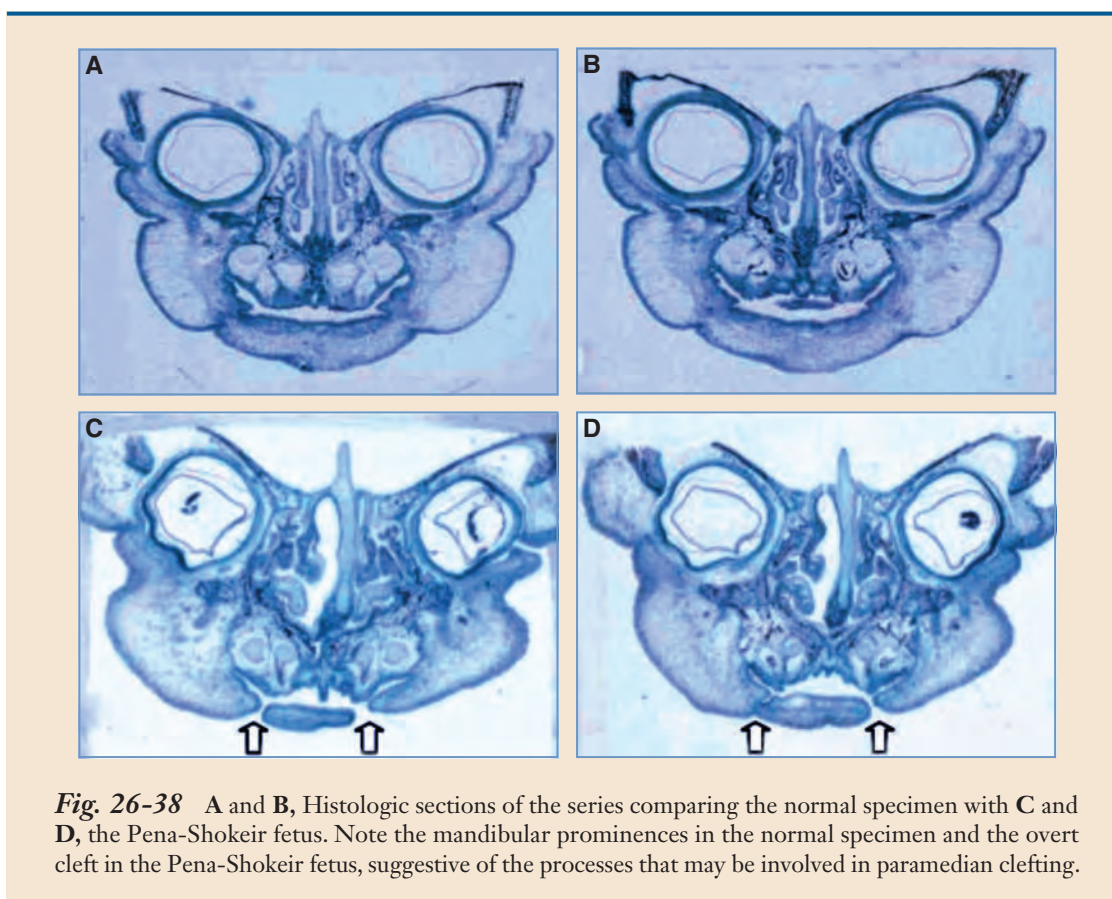
notch in the vermillion to a true median cleft with involvement of the entire vertical length of the soft tissue¹⁴² (Fig. 26-36). Varying degrees of involvement of the tongue have been reported. The tongue may be absent or more commonly bifid. The medial margins of the bifid tongue may be bound down to the margins of the cleft. The complete duplication of the tongue is at the extreme. The cleft in the mandibular alveolus begins between the central incisors and extends to involve the mandibular symphysis.¹³⁵⁻¹⁴²

As the median cleft continues caudally to involve the lower branchial arches, the structures derived from them are involved. The hyoid bone, thyroid cartilage, and sternum may be clefted, and the associated musculature may be hypoplastic. A congenital neck cord may be present as a pathognomic sign of the underlying cleft.¹⁴³⁻¹⁴⁷



Tessier 28/29 Cleft: Paramedian Mandibular Cleft

Tessier did not formally designate a number for the paramedian mandibular clefts, probably because of its rarity. He may have also decided to leave the classification scheme open until there were sufficient clinical cases available for an orderly scheme. Only a handful of cases have been documented in the literature within the past century. Among the earliest is the case in 1952 of a newborn with ankyloglossia and symmetrical bilateral clefts of the lower lip.¹⁴⁸ However, the three additional cases in the literature have been a unilateral paramedian mandibular cleft and associated with varying degrees of hemifacial microsomia and Goldenhar syndrome.¹⁴⁹⁻¹⁵² Within the existing Tessier nomenclature, these rare paramedian clefts would be numbered between Tessier 14 and 30 clefts. Morritt et al¹⁵¹ suggest a paramedian cleft of the lower lip in the case they present as a Tessier 28/29 (Fig. 26-37).



Although the median mandibular cleft can be simplistically understood as failure of fusion of the paired first branchial arches, the existence of paramedian clefts then belies an explanation. There must be another process at the embryologic level that accounts for both the median and paramedian clefts. In their detailed study of the Pena-Shokeir fetus, Vastardis et al¹⁵³ found a set of paired paramedian surface furrows that separated the central mandibular region from the lateral elements (10 to 16 mm CR length, day 38). Thus failure of growth center differentiation could account for the contribution to the bilateral and unilateral forms of mandibular paramedian clefts. Therefore the median cleft could be considered a bilateral form without the central portion of the symphysis (Fig. 26-38).

MANAGEMENT OF FACIAL CLEFTS

General Principles

Given the rarity and unique nature of the clinical expression of each of the craniofacial clefts, the treatment plans cannot be standardized but must be based on the individual assessment of each case (Figs. 26-39 through 26-41). Nevertheless, there are general principles by which the sequence and timing of staged osseous and soft tissue reconstruction can proceed.

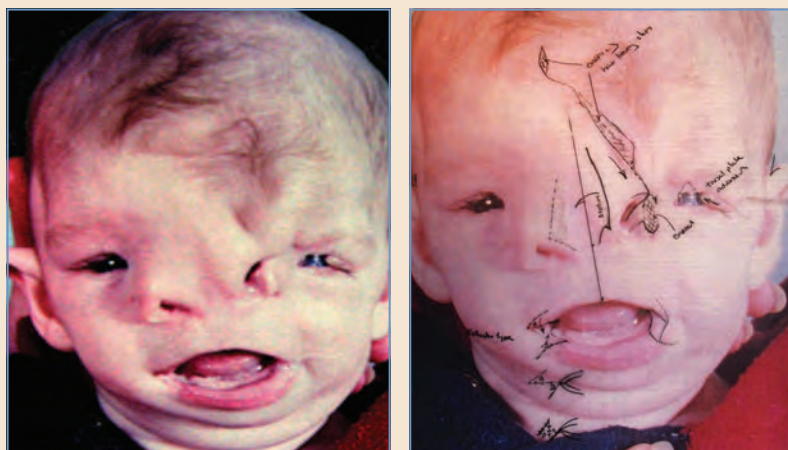


Fig. 26-39 In more complex clefts, reconstruction requires staging the soft tissue and skeletal components based on potential treatment of vital structures and growth development of the child. In infants reconstruction involves repairing the upper eyelid defect with a tarsal advancement flap, initial nasal reconstruction with inferior advancement of the nasal dorsal tissue, and macrostomia repair. The correction of the bifid nose and hypertelorism is delayed until later in childhood.

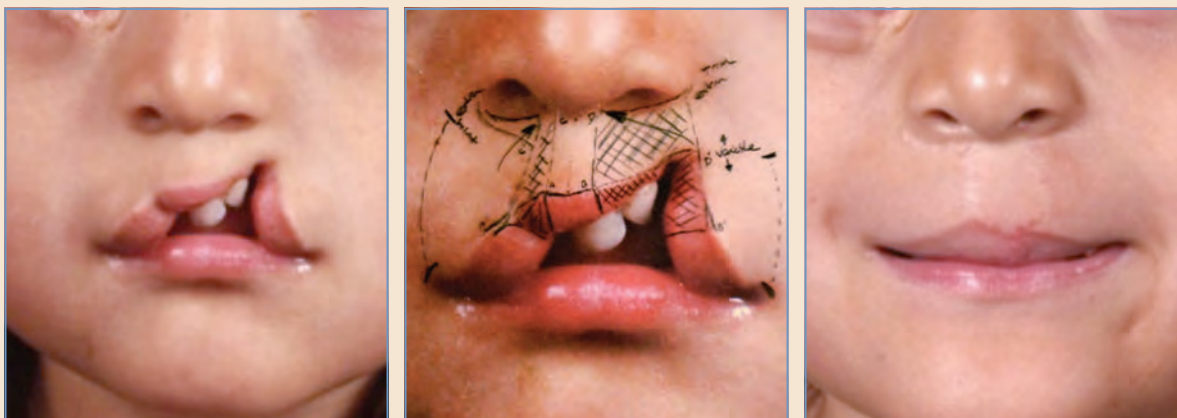


Fig. 26-40 Atypical cleft lip reconstruction. Overlaying cephalometric tracing paper is useful for planning. Right lower lid coloboma reconstruction is planned.



Fig. 26-41 Complex atypical craniofacial cleft reconstruction that required tissue expansion.

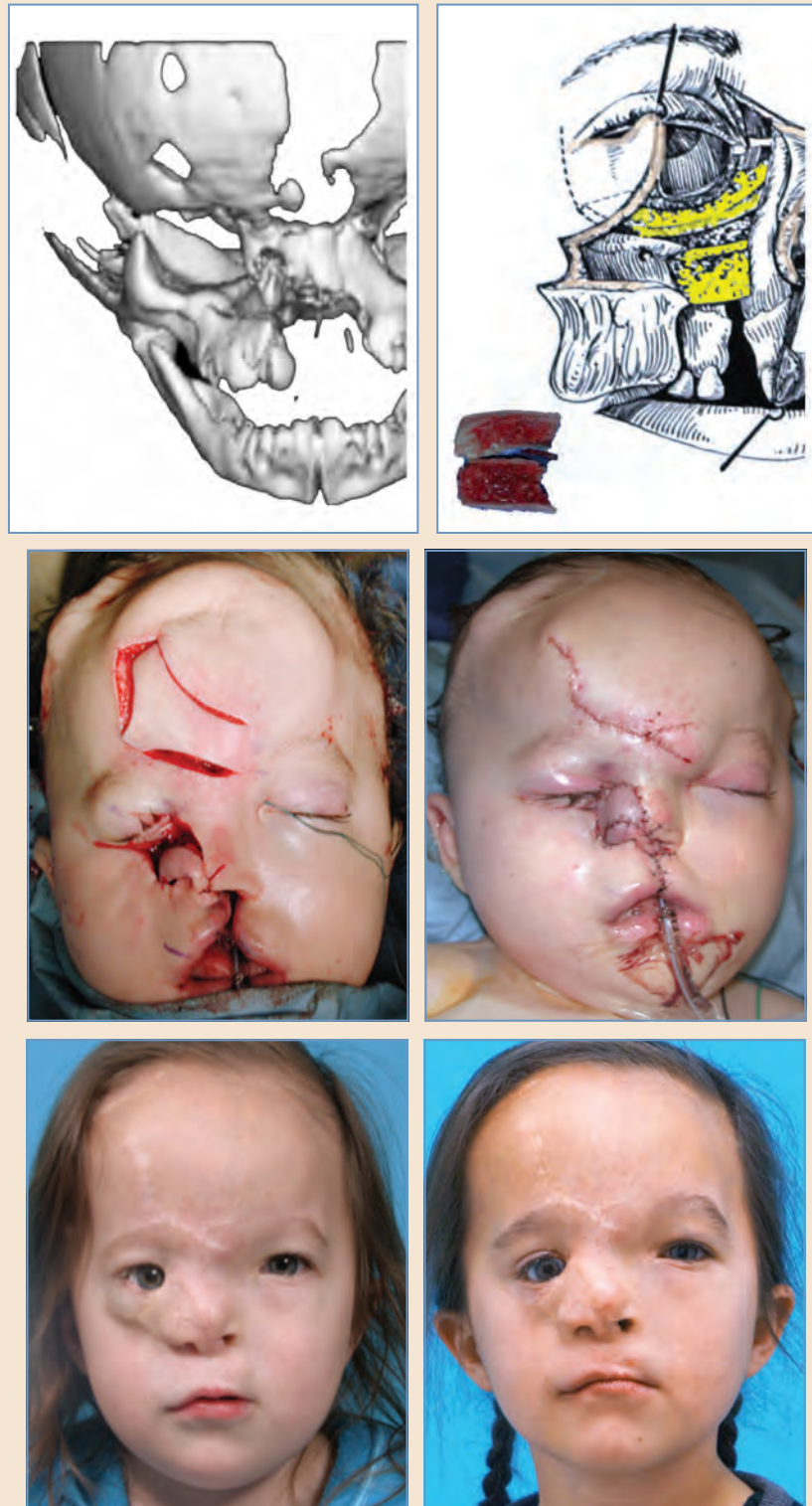


Fig. 26-41, cont'd At initial surgery, the skeletal defect was reconstructed with rib grafts to provide support for the soft tissue flaps. The cleft palate was reconstructed simultaneously. Further reconstruction is planned.

The defect can be corrected with a single operative procedure only in the mildest expression of the facial cleft. As such, most of the cases that the surgeon encounters will require careful staging of the reconstruction. The two questions that the surgeon must answer include (1) what should be the sequence of skeletal and soft tissue reconstruction based on the relative inventory of the involved anatomic structures, and (2) what are the consequences of each of the staged procedures and the durability of the reconstruction with continued growth of the child?

The severity of the malformation in terms of a potential threat to a vital structure, such as the eye, dictates an early intervention. The initial reconstruction should begin as soon as the infant is physiologically able to withstand anesthesia and the surgical insult. When the actual cleft deformity does not pose a significant risk, an appropriate delay can take advantage of the growth of the child, with an appreciable increase in the size of the component structures that facilitate accurate alignment and appropriate positioning of the key landmarks.^{10,154,155}

The initial operative intervention is generally limited to the soft tissue structures. Even a minimal crease as an expression of a facial cleft may involve all underlying layers, including subcutaneous tissue and muscle. The intervening abnormal tissue within the region of the cleft must be excised to expose the normal structures and then accurately aligned to reconstruct the anatomy. Failure to meticulously restore the continuity across the cleft can result in a loss of muscular animation or may leave a persistent stigmata, such as a depressed scar, because of the lack of appropriate soft tissue support. When the repair crosses the lines of minimal tension, the closure should be redirected to a more favorable position with multiple Z-plasties. When there is a difference in the length of the vertical height between the two sides of the cleft, carefully planned rotation/transposition flaps, along with multiple Z-plasties, can be used to restore the length and proper position of the anatomic landmarks on the face. In designing the regional flaps, it is important to remember the soft tissue facial units, and that the scars are best camouflaged by the natural folds and creases to avoid a patchwork quilt appearance to the soft tissue repair.^{8,10,120,155,156}

When the regional tissue is considered insufficient to correct with local flaps alone, an alternative approach is to use tissue expanders to restore the soft tissue defect with adjacent expanded skin of similar color, texture, and appearance. Expanding the soft tissue envelope before skeletal restoration may be beneficial. The success of a bone graft depends on adequate soft tissue vascularity and an envelope free of tension. Furthermore, in later stages, expansion of the regional tissue may allow excision of grossly scarred regions from multiple previous operations and resurfacing with soft tissue of improved color, texture, and vascularity.¹⁵⁷

Although repair of the soft tissue deformity can splint the bony discontinuity and potentially mold the underlying distorted facial skeleton without proper restoration of the structural skeletal support to the soft tissue reconstruction, the stigmata of a facial cleft will always remain. Moreover, a maxillary alveolar cleft creates structural instability of the midface and segmental collapse. The time to optimally intervene remains unsettled. Early manipulation of the underlying bony skeleton in the region may potentially disturb normal midface development. Bone grafting the cleft defect early may lock the maxillary segments in place and interfere with the normal direction of growth and remodeling. Although arguably the inherent growth potential in these patients remains unknown and may possibly be limited because of the severity of the anomaly, early restoration of the skeletal contour can significantly lessen the deformity and allow the child to be “mainstreamed” earlier into society. However, what may be an acceptable result at age 5 or 6 years may be less than ideal in the long-term.^{10,126,158,159}

Although the cranial vault grows rapidly in the first year of life, it levels off earlier than the rest of facial development. The upper midface grows at a slower rate in infancy, but it continues to grow later in childhood and early adolescence. In contrast, growth of the lower midface and mandible, which is determined by eruption of the dentition, continues well into adolescence. The overall growth of the cranioorbitozygomatic regions reaches approximately 85% of its adult size by 5 years of age.¹⁶⁰ Consequently, skeletal reconstructive surgery involving the upper midface

(cranioorbitozygomatic region) should be relatively stable after 5 to 6 years of age. In contrast, reconstruction of the maxillary-mandibular complex should be delayed until adolescence, although there are exceptions. Unless significant functional concerns arise, the timing of reconstruction should be based on an understanding of the normative residual growth expected within the operated region of the facial skeleton.¹⁰

Fundamental principles of plastic surgery procedures apply to all facial clefts. However, specific considerations arise as the individual clefts are addressed.

Number 0-14 Clefts

The true median cleft of the upper lip is repaired by excision of its margins, approximating the muscle layer and accurately aligning the vermilion border to restore the philtral width. The frenulum should be excised when it can result in a diastema of the central incisors. Correction of the bifid nose requires excision of the central furrow and medial displacement of the alar cartilage and nasal bone osteotomies. A dorsal bone graft (cranial or rib) may be required for structural support and increasing the anterior projection of the nasal tip.^{10,110,161}

When there is an associated hypertelorism, a facial bipartition should be considered to simultaneously correct the interorbital width, nasal deformity, and centrally collapsed dental alveolar arch. Depending on the extent of the interorbital involvement and level of the cribriform plate, either an intracranial or subcranial approach is used. Osteotomies are made to mobilize the hemiface. Laterally, the zygomatic arch is cut, and the osteotomies of the lateral orbital wall are extended to the pterygomaxillary junction. Medially, the osteotomy of the palate and premaxilla is completed, and the central mass of the widened nasal skeleton and ethmoid is resected to correct the interorbital width. As the two halves of the face are mobilized medially, the central part of the face is elongated. The palpebral angulation is improved with rotation of the orbit, and the anterior open bite is corrected with the dental arch leveled and the transverse palatal width increased.*

At the other end of the spectrum of the 0-14 clefts are the cases of holoprosencephaly-related malformations, characterized by central hypoplasia. Infants with cyclopia, ethmocephaly, or cebocephaly (groups I through III of the DeMyer et al⁸⁰ and Elias et al⁹⁰ classifications) do not survive infancy and surgical intervention would be inappropriate. In contrast, surgical reconstruction should be offered to infants with less severe neurologic compromise. Infants who fall into group IV (lobar brain morphology) are likely to survive, and repair of the false median cleft lip and palate could be offered. Because these children are invariably mentally retarded, more extensive procedures would be of little benefit. In contrast, within the group of children with grossly normal brain anatomy (group V), a subcranial LeFort III with facial bipartition can be offered to correct the hypotelorism, midface retrusion, and transverse maxillary arch deficiency.^{90,115}

Clefts 1 and 2

Similar to the familiar cleft lip and palatal deformity, the cleft in the Cupid's bow region can be repaired by well-described rotation-advancement techniques. The correction of the alar deformity of the first and second cleft may require either cartilage or composite grafts to the alar rim. More significant disruptions require a complex design of internasal lining and overlying cutaneous flaps with cartilaginous support. The dentoalveolar segment will require bone grafting at the time of mixed dentition coordinated with orthodontic treatment.†

*References 10, 18, 111, 154, 155, 162.

†References 8, 10, 155, 156, 163, 164.

Clefts 3, 4, and 5

Cleft 3

Although the cleft of the lip begins in the Cupid's bow region, it disrupts the alar base and medial canthal region. As a result of the discontinuity, the vertical dimension of the soft tissue between the medial canthus and ala is foreshortened, with the ala rotated cephalad and the medial canthus drawn caudally. Restoring the symmetry of the elements and proper vertical dimension may require the use of an asymmetrical Z-plasty or rotation/transposition flaps with extensive undermining of the soft tissue of the cheek. The medial canthal ligament is hypoplastic, and repositioning the canthus superiorly medial to an anatomic position requires the use of transnasal wiring (Fig. 26-42). As a result of the path that the cleft takes, the nasolacrimal system is disrupted and prone

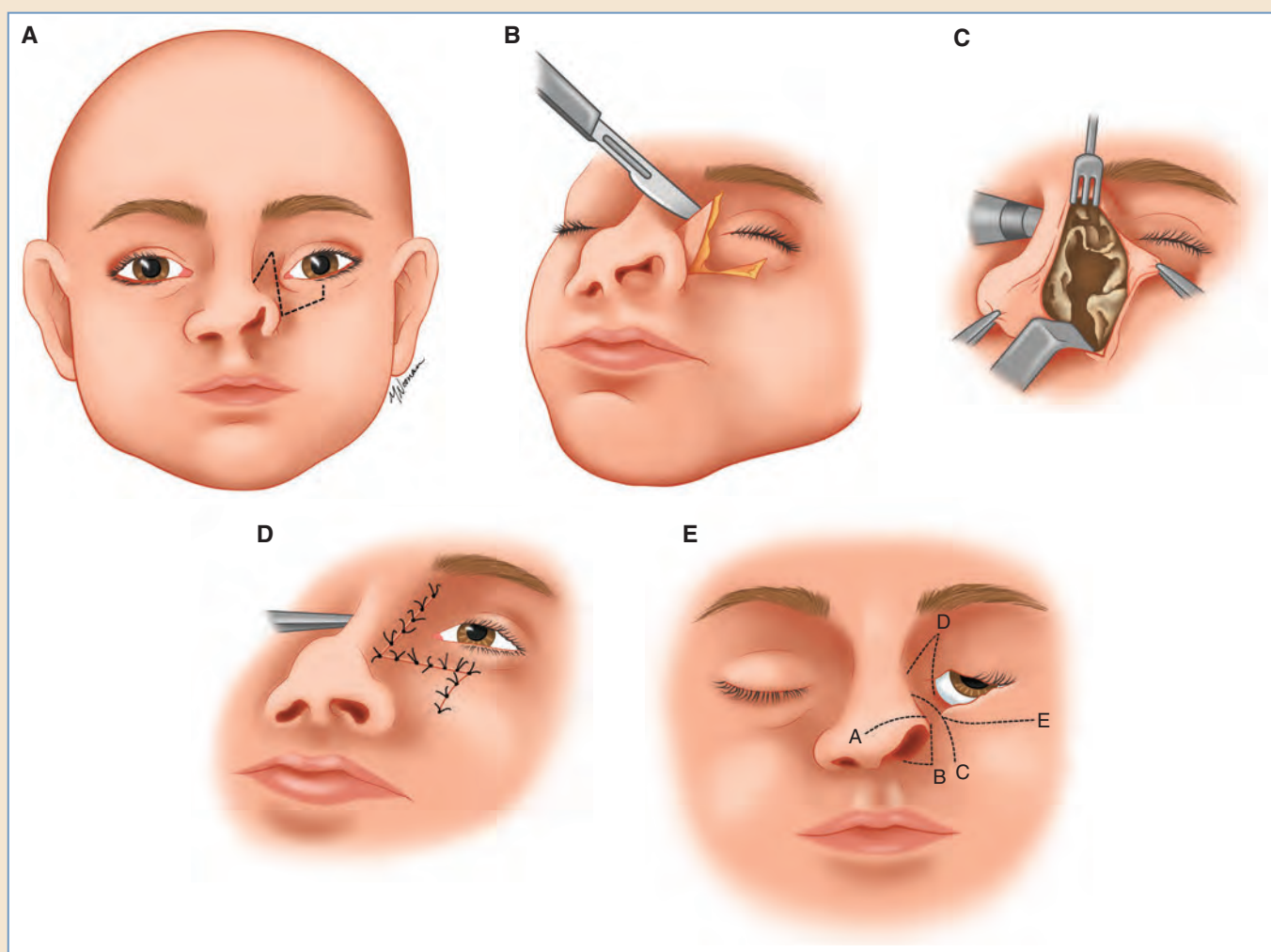


Fig. 26-42 The number 3 cleft repair. **A-D**, Transposition Z-plasty flaps are used to reposition the medial canthus superiorly and medially while simultaneously lowering the raised ala. Elevation of the transposition flaps, transnasal medial canthopexy, and closure after bone grafting of the underlying skeletal defect are shown. **E**, A single transposition flap may be insufficient to correct the cleft ala. Each cleft is unique and tests the ingenuity of the surgeon to design various local regional flaps to reconstruct the tissue deficiency.

to repeated infections. Often beyond repair, the most satisfactory result is obtained by complete removal of the sac, duct, and canalicular system during the initial soft tissue reconstruction.

In the complete form, the normal nasal maxillary septation is absent, and the oral, nasal, maxillary sinus, and orbital contents are in continuity. The orbital volume and contour of the inferomedial orbital rim is restored with bone grafts, with mucosal lining obtained from adjacent nasal and maxillary mucosa. Bone grafting of the dentoalveolar segment and anterior maxillary wall to support the perialar region should await the stage of mixed dentition.*

Cleft 4

With the cleft of the lip occurring lateral to the philtrum, the lip repair requires discarding the entire labial tissue between the philtral column to the cleft edge. Failure to excise the tissue seems to result in a less than optimal result with the orientation of the scar, persistent appearance of macrostomia, and deficiency of the orbicularis oris muscle in the cleft region (Fig. 26-43).

*References 8, 10, 47, 117, 120, 155, 156, 165, 166.

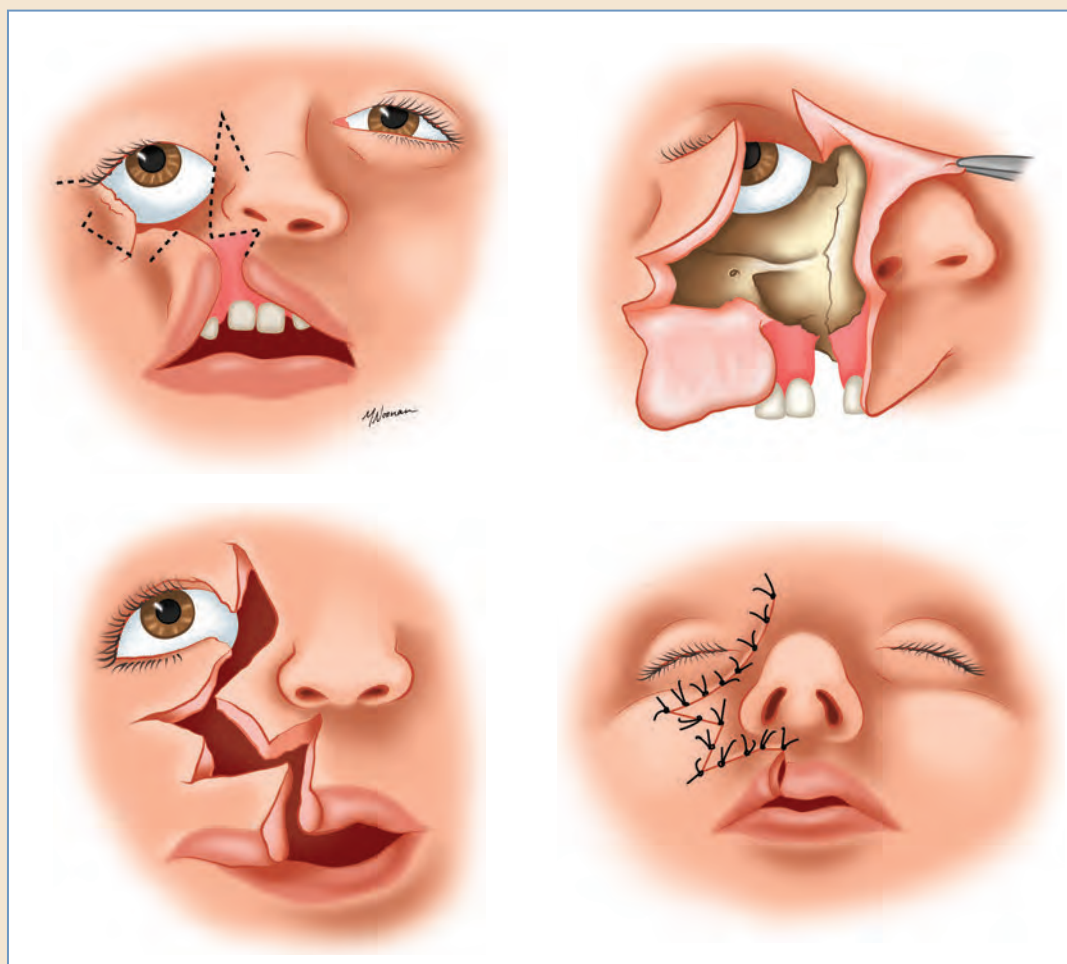


Fig. 26-43 The number 4 cleft repair. Triangular and quadrilateral transposition flaps are used to restore the vertical dimension and anatomic features. The medial canthus and ala are repositioned. The orbital floor and maxillary defects are bone grafted.

Although the cleft spares the nose and piriform aperture, the ala on the affected side is rotated cephalad, as is the lateral lip element with the deficiency of soft tissue. The decreased vertical height medially is corrected by allowing both the ala and lateral lip element to rotate caudally. This will require wide undermining of the soft tissue and careful design of the interdigitating flaps (see Fig. 26-40).

With the remaining portion of the nasolacrimal system functioning, the disrupted inferior canaliculus is left unreconstructed (see Fig. 26-43). The medial canthal tendon is intact and can be used to anchor the tarsal plate of the lower lid as it is repositioned medially and superiorly to help protect the globe. The deficiency in the conjunctival fornix may require a mucosal graft. Correction of the orbital dystopia and position of the ocular globe requires bone grafting the defect in the medial orbital rim and floor. The defect in the anterior maxillary wall is grafted to support the overlying soft tissue. Bone grafting of the dentoalveolar segment of the cleft should wait for the stage of mixed dentition, coordinated with orthodontic treatment.*

Cleft 5

Like the number 3 and 4 clefts, soft tissue reconstruction of the number 5 cleft involves rotation/transposition flaps to correct the vertical deficiency. Although the soft tissue approximation is simplified without involvement of the medial canthal region, the obliquity of the scar from the region of the commissure to the lateral portion of the lower eyelid is unfavorable. The interdigitation of the flaps requires respecting the facial units and redirecting the limbs along what will eventually be the natural folds and creases. Repair of the cleft of the upper lip should lie along the nasolabial crease.

Restoration of the skeletal defect is similarly carried out by bone grafting the lateral third of the orbital rim and floor to restore the support to the ocular globe. The dentoalveolar defect in the anterolateral maxillary region will also require bone grafting.^{10,97,155}

Clefts 6, 7, and 8

Cleft 6

The Tessier cleft 6 is usually mild. The soft tissue reconstruction involves correcting the antimonogloid slant of the palpebral fissure and restoring the continuity of the lower eyelid coloboma. The skeletal defect requires bony augmentation of the hypoplastic zygoma to restore the malar height and supporting the globe with bone grafts to the lateral orbital floor and rim. The discontinuity of the lateral zygomaticomaxillary buttress should also be bone grafted.^{10,155,167}

Cleft 7

The number 7 cleft has a broad spectrum of severity, and its reconstruction is tailored to the patient. Treatment planning is complex and various with the age at presentation and degree of soft tissue and skeletal involvement. Most commonly, small preauricular tags may represent the mildest form of the cleft and can be removed early in infancy. Patients with the more severe auricular deformity of microtia can begin reconstruction at 5 or 6 years of age when appropriate. Depending on the degree of auricular dystopia, this may require repositioning the auricular remnant posterior-superiorly and the use of a temporoparietal fascial flap. The auricular reconstruction is described in Chapter 31. The macrostomia seen as a transverse cleft or deep furrow should be repaired early. The margins of the cleft should be excised, the orbicularis muscle identified, and the muscle fibers reoriented to restore the continuity of the circumoral musculature. The technique has been well described in the literature by May,¹⁶⁸ Stark and Saunders,¹⁶⁹ Boo-Chai,¹⁷⁰

*References 10, 18, 47, 97, 126, 155, 164, 165.

Bauer et al,¹⁷¹ and Verheyden.¹⁷² The generalized loss of soft tissue bulk (subcutaneous and muscle) requires the use of either serial dermal fat grafts because of the unpredictability of tissue resorption or a vascularized free tissue (for example, deepithelialized scapular or groin) transfer.^{173,174}

Reconstruction of the skeletal deficiency is dependent on the degree of maxillary and mandibular involvement and the resulting asymmetry and occlusal relationship. Reconstruction may require restoring the continuity of the zygomatic arch, mandibular lengthening (either distraction osteogenesis or conventional rib grafting) at the stage of mixed dentition, and maxillary-mandibular osteotomy when facial growth is complete to correct the end-stage asymmetry.* The treatment planning is described in Chapter 31.

Cleft 8

The soft tissue reconstruction of the number 8 cleft involves reconstructing the normally sharp angle of the lateral canthus by restoring the lateral canthal ligament, depth of the conjunctival fornix, length of the palpebral fissure, and continuity of the oculi muscle. Although direct closure of the lateral canthal defects has been described, the technique of Fuente del Campo¹⁷⁸ is an ideal choice, because it restores the normal form to the region. Flaps are created with the superior and inferior palpebral folds and the intervening tissue in the coloboma. The fibrous tissue of the hypoplastic lateral canthal ligament is then fixed to the orbital rim at the appropriate level. The orbicularis muscle is dissected and approximated. The superior and inferior palpebral flaps are approximated, defining the palpebral width, and the laterally based flap fills the resulting superior and inferior defects.

The bony defect in the lateral wall of the orbit is usually insignificant and is restored when orbicularis oculi muscle is approximated. In rare cases, split calvarial bone may be required in more significant defects.^{10,155,167}

Clefts 6, 7, and 8—Treacher Collins Syndrome

The pseudocoloboma of the lower lid can be improved with either a Z-plasty or an upper-to-lower lid transfer and could be corrected before reconstruction of the orbitozygomatic complex and lateral canthopexy.^{167,179} The auricular reconstruction typically begins at about 6 years of age and is outlined in Chapter 31.

The reconstruction of the Treacher Collins syndrome (bilateral cleft 6, 7, and 8 combination) primarily depends on correcting the underlying skeletal architecture. The deficiency in the lateral orbital wall, anterior floor of the orbit, and inferior orbital rim requires bone grafting to restore the orbital morphology. The antimongoloid slant is corrected by repositioning the lateral canthus and contouring the superolateral orbital slant. Lateral canthoplasty must be performed each time the orbital complex is regrafted, and there is an invariable downward drift despite fixation. The continuity of the zygomatic arch should be reestablished, and additional onlay bone grafts should be done to restore the anterior projection of the hypoplastic malar region. In harvesting the required amount of cranial bone, care must be taken not to injure the vascular supply to the temporoparietal fascia, which may be used to support the skin graft in the auricular reconstruction. The amount of bone required is enormous, and skeletal reconstruction should be delayed until the child is older than 6 years of age. Moreover, as Tessier⁹⁸ has noted, resorption is more severe in Treacher Collins syndrome than in other malformations and requires repeated bone grafts to maintain the bony volume.

To improve the dental relationship and facial balance, maxillary and mandibular procedures are required. Correction of the vertically short posterior maxilla may be achieved by a LeFort I

*References 10, 155, 167, 173, 174-177.

osteotomy or require a LeFort II osteotomy with rotation (not advancement) to lower the maxillary tuberosity and simultaneously enlarge the nasopharyngeal space. To correct class II malocclusion and the anterior open bite deformity, exaggerated even more with the maxillary movement, the mandible is lengthened and advanced. Although a sagittal split osteotomy is possible in some patients, the ramus is often short and narrow, and a V osteotomy with bone grafts is preferred. With this approach, Tessier⁹⁸ describes mandibular advancements of 20 to 25 mm and vertical lengthening of the ramus without practical limits. In addition, osseous advancement genioplasty with elongation of the suprahyoid tissues improves the contour.

When respiratory insufficiency occurs as result of the narrow nasopharyngeal/hypopharyngeal space, surgical intervention may be required. Unlike the adult, the midface in the child must be rotated at the level of a subcranial LeFort III to avoid the developing tooth buds. Recently, the introduction of mandibular osteotomy and distraction osteogenesis may play a role in these cases of children who require a tracheostomy.^{10,167,177,180-182}

Clefts 9, 10, and 11

The soft tissue reconstruction of the number 9, 10, and 11 clefts involves restoring the discontinuity in the upper eyelid and brow with adjacent tissue transfer. The bony cleft in the superior orbital rim and roof requires bone grafting, and the associated deformity in the frontal bone should be augmented to restore the proper projection to the supraorbital region of the brow.

In severe cases of clefts 10 and 11, orbital dystopia and hypertelorism can result and require both an intracranial and extracranial approach.^{10,155,183,184}

Clefts 12, 13, and 14

The reconstructive efforts in the number 12, 13, and 14 clefts are primarily directed to correcting the hypertelorism that results from widening the ethmoidal labyrinth in the number 12 cleft and the cribriform plate, beginning with the number 13 cleft. Both clefts 13 and 14 can present with an encephalocele that must be addressed with both an intracranial and extracranial approach.^{10,155,183} Surgical correction of hypertelorism is discussed in Chapter 25.

Cleft 30

Reconstruction of the mandibular symphyseal cleft involves restoring the continuity of the mandibular skeletal symphysis by approximating and fixation of the mandibular cleft border. Reconstruction of the overlying cleft soft tissue involves restoring the continuity of the orbicularis muscle, midline cervical muscles, and overlying skin with appropriate transposition and Z-plasty flaps. The surgical technique is well described in several publications.¹⁸⁵⁻¹⁸⁷

CONCLUSION

The laws of normal growth are best formulated and understood when the causes of their exception can be established. The experimental method itself rests on the notion that induced and controlled departures from the ordinary can unveil the laws of order. Consequently, congenital malformations are nature's experiment, uncontrolled by intentional human art to be sure, but source of insight nevertheless. The early teratologists sought to understand malformations by classifying them. The early French anatomists developed three categories: missing parts (*monstres par défaut*), extraparts (*monstres par excès*), and normal parts in the wrong places. However, a classification is no more than a set of pigeon holes until the causes of the ordering can be specified.

KEY POINTS

- Atypical facial clefts are 100 times less common than common clefts.
- Radiation, infectious agents, metabolic imbalances, and numerous drugs and chemicals have been implicated as causes of atypical craniofacial clefts.
- Extrinsic mechanical factors, such as from an amniotic band disruption mechanism, have been suggested to explain aberrant locations of clefts not corresponding to any well-described embryologic process.
- Historically, two theories of facial clefting have been described: the classic theory of fusion by Dursy⁵¹ and His⁵² and the theory of mesodermal migration by Veau and Politzer,⁵³ Warbrick,⁵⁴ Stark,⁵⁵ and others. Other more recent theories proposed by Johnston⁵⁶ and Vermeij-Keers et al⁵⁷ have led to possible explanations of some of the rarer defects clinically seen.
- Studies by Vermeij-Keers et al⁵⁷ and others have shown that migration of neural crest cells alone may not account for all varieties of facial clefting. Rather, facial clefts may represent failure of local differentiation of the neuroectoderm and mesenchyme. They identified four primary sites in early embryologic development that could lead to true cleft formation. They are associated with the normal planes of embryologic processes, and their closure occurs sometime within the seventh gestational week.
- After the seventh gestational week, the differentiation of the mesenchyme into facial muscles and supporting bone and cartilaginous support occurs. Ossification centers arise to complete the bone formation. If the ossification centers fail, then pseudoclefts develop. These are not true clefts, and as such, have been termed *secondary clefts or dysplasias*.
- Holoprosencephaly is defined as a spectrum of median facial deformities associated with tissue-deficient median cerebrofacial dysmorphogenesis.
- The various degrees of facial dysmorphism associated with the severity of holoprosencephaly have been grouped into five major types: (I) Cyclopia, (II) ethmocephaly, (III) cebocephaly, (IV) hypotelorism with median cleft lip, and (V) hypotelorism with bilateral cleft lip.
- The degree of correlation between the brain and face is not absolute. The predictive correlation is only about 80%. The scheme of DeMyer et al⁸⁰ has been revised by Elias et al⁹⁰ to reflect those variations of clefting, in which the brain is either near normal or normal despite the facial clefting.

Continued

KEY POINTS (continued)

- DeMyer⁹¹ proposed the term *median cleft face syndrome* to describe this collection of median facial anomalies characterized by near-normal or excess tissue. The dysmorphic features can vary from a subtle midline notch of the upper lip to an obvious bifidity of the nose and in more severe cases hypertelorism. These defects are believed to occur late—after the seventh week of gestation.
- Numerous classification schemes have been proposed for rare atypical clefts. Some are based on the embryology, whereas others are based on the clinical findings. Although a classification scheme, including both a descriptive terminology and insights into the underlying embryology, is preferable, Tessier's system⁹⁸ has proved the most useful clinically.
- In 1976 Tessier's classification system⁹⁸ represented an architectural description of defects rather than true descriptions of the structures involved. The defects are numbered for ease of clinical classification rather than along any pathoembryogenic sequence. Although it fails to provide insight into the underlying etiopathology of these defects, the relative simplicity of this classification has made it almost universally accepted for clinical use.
- The most common cleft types seen are the median clefts 0 and 14, although the transverse cleft, number 7, would be expected to be significantly more common with a stricter definition.
- Careful observation of the atypical cleft anatomy, coupled with a clear understanding of the normal and abnormal embryologic processes that affect each structure, provide both the basis of classification and keys to reconstruction.

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27

Orthognathic Surgery in Adolescents With Dentofacial Skeletal Deformities

Pravin K. Patel • Stephen B. Baker



Orthognathic surgery involves the surgical manipulation of the elements of the facial skeleton to restore the normal anatomic and functional relationship in patients with anomalies of the dentofacial skeleton. This chapter will provide an overview of the principles of orthognathic surgery that can be used to manage a broad spectrum of maxillofacial skeletal abnormalities, whether of congenital, developmental, or acquired origin.

The word *orthognathic* comes from the Greek *orthos*, meaning “straight,” and *gnathos*, meaning “jaw”; thus orthognathic surgery means to straighten a jaw. Differentiating a straight jaw from one that is not requires that the surgeon determine the degree of deviation from a specified population norm. Ultimately, restoring the orthognathic form of the face depends on achieving the ideal facial aesthetic for the individual patient and not simply on restoring the average normative values of any given population.

Of course, the face encompasses more than the upper and lower jaws, and when deformities also involve the cranioorbital skeleton, evaluation and management expand the scope of maxillofacial surgery to include craniofacial surgery. Thus orthognathic or maxillofacial surgery should be considered a subcategory of craniofacial surgery.¹

In general, dentofacial skeletal anomalies occur because of a difference in growth between the upper facial skeleton and lower facial skeleton, resulting in a discrepancy in the normal relationship that exists between the upper and lower jaws. The normal growth of the facial skeleton can

be influenced by an underlying genetic predisposition and acquired causes. Congenital anomalies from facial clefts resulting from various syndromic conditions such as Apert and Crouzon syndromes affect normal growth and development. Traumatic events in the mature skeleton can displace the normal elements and require repositioning osteotomies if they are improperly reduced initially, or in the developing facial skeleton, they can interfere with normal subsequent growth. However, the developmental anomalies representing the extremes of population norms are the most common conditions requiring orthognathic surgery.²

Indications for orthognathic surgery include facial dysmorphism with or without functional implications. For example, in patients with retrogenia but without malocclusion, an osseous genioplasty might be considered for facial form. If the retrogenia is associated with retrognathism resulting in malocclusion, then orthognathic surgery is used to restore not only the facial form but also functional occlusion. From a functional standpoint, the goal of orthognathic surgery is to reestablish the following equation:

$$\text{Centric relation} = \text{Centric occlusion} = \text{Habitual occlusion}$$

Thus both function and appearance must be optimized from multiple perspectives.

Maxillofacial deformities can be broadly divided into three major categories: (1) skeletal dysplasias, (2) dental dysplasias, and (3) dentofacial skeletal dysplasias. Dental dysplasias are strictly limited to malocclusions that result from an abnormal spatial relationship of the dentition alone and not because of the skeletal position of the upper and lower jaws. These can be corrected with orthodontic treatment alone. In patients with skeletal dysplasia alone, the dentition is in good alignment, but the maxilla and/or mandible is dysplastic. Skeletal dysplasias require correction of the skeletal deformity without altering the occlusion (for example, in a patient with retrogenia without retrognathia or one with hemifacial microsomia who has a normal maxillary-mandibular dental relationship but an occlusal cant because of skeletal asymmetry). In dentoskeletal dysplasias, the dentition is malpositioned within each arch and between the upper and lower arches; in addition, the skeletal relationship between the upper and lower jaws is abnormal. An example is a patient with a maxillary sagittal and transverse width deficiency resulting from a facial cleft. Correction requires aligning the dentition within each arch with orthodontic treatment and restoring the maxillary-mandibular dental relationship with skeletal osteotomies and repositioning.

Skeletal and dentofacial skeletal dysplasias can be further classified based on the position in space and the volume or mass (whether deficient or excess) of the individual facial elements. For example, the mandible can be of normal shape and volume but retrognathic in relation to the maxilla, or it may be not only retrognathic but also volumetrically deficient. Each of the elements—the zygoma, maxilla, mandible, and chin—can individually be disproportionately affected, leading to a variety of clinical presentations (Fig. 27-1). However, similar clinical appearances can occur as a result of very different structural problems, and determining which facial elements are involved in the deformity requires a systematic evaluation (Fig. 27-2).

EVALUATION OF DENTOFACIAL DEFORMITIES

Correction of maxillofacial deformities requires careful analysis of the soft tissue with clinical examination and supporting photographs, skeletal evaluation with standardized radiographs, and dental evaluation with study models.³⁻⁶ This comprehensive evaluation from multiple perspectives will then generate a list of problems, all of which will need to be addressed to achieve the intended goal.



Fig. 27-1 Diversity of dentofacial skeletal deformities. **A** and **B**, Midfacial deficiency (maxillary retrognathia/hypoplasia) in a patient with a facial cleft. **C** and **D**, Mandibular deficiency (retrognathia/hypoplasia) of developmental origin. **E** and **F**, Vertical maxillary excess with mandibular retrognathia and retrogenia of developmental origin. **G** and **H**, Maxillary-mandibular asymmetrical growth in a patient with hemifacial microsomia.

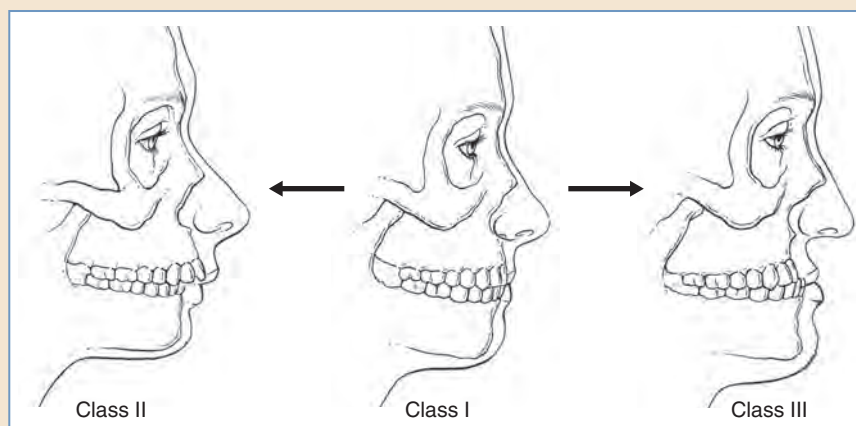


Fig. 27-2 Range of dentofacial deformities. Class I (center) orthognathic deformity. Class II (left) dentofacial skeletal deformity. Sagittal maxillary excess and mandibular retrognathia can result in similar appearances. Class III (right) dentofacial skeletal deformity. Sagittal maxillary deficiency and mandibular excess can result in similar appearances.



Fig. 27-3 A, Frequently patients will hold their lips together to avoid showing their teeth. This results in mentalis muscle strain. B and C, The lips must be in a relaxed position to clinically assess the lower third facial structures.



Fig. 27-4 Range of variations in the position of the lips and the amount of dental display. A, Lips together; B, relaxed lip position; C, natural smile; and D, broad smile. Ideally, patients should be assessed with the soft tissues (facial muscles) in a relaxed position and the lips in repose. E-H, Patients should be assessed in frontal, lateral, and oblique views with the head in the Frankfort horizontal plane and the eyes parallel with the horizon.

Clinical Assessment

Clinical assessment should be directed specifically at evaluating the degree of zygomatic projection, the maxillary and mandibular positions in space relative to each other and to the cranial vault, and the relative size or proportion of each of the facial elements. However, proper orientation of the head with respect to the horizon is important in determining which elements of the facial skeleton need to be repositioned. Patients with dentofacial deformities may alter their head posture to improve function or make the deformity less noticeable, and it becomes important to adjust for such compensations at the time of clinical, photographic, and radiographic examinations.⁷

Patients should be seated upright and asked to look toward the horizon. The head should be oriented with the clinical Frankfort horizontal plane (the line from the ear tragus to the infraorbital rim) parallel to the floor. Equally, the posture of the lips needs to be standardized.^{8,9} Frequently patients will unconsciously force their lips together to avoid showing too much of their teeth, which results in straining of the mentalis muscle and also affects the chin contour (Fig. 27-3). When patients are being examined, the lips should be in a relaxed position so that there is a natural draping of the soft tissue. With relaxed lip posture, the degree of dental display (lip-to-tooth ratio), evidence of lip incompetence, dental-facial midline discrepancies, and, when combined with relaxation of the mentalis muscle, chin contour can be determined. With the head properly positioned, the head mimics the natural standing posture and helps to standardize the measurements, treatment planning, and assessment of the outcome after orthodontic-surgical treatment.

Examination of the face should be systematic, with assessment of the frontal, profile (lateral), and three-quarter (oblique) views¹⁰⁻¹² (Figs. 27-4 and 27-5). The frontal analysis should focus on recording left-right facial symmetry, the presence or absence of the occlusal cant in the frontal plane, the congruence of the midline structures between the maxillary-mandibular dental midline with the facial midline, the length and posture of the upper lip, and the amount of dental display with the lips in repose and with a natural smile.^{13,14} Examination of the patient in profile

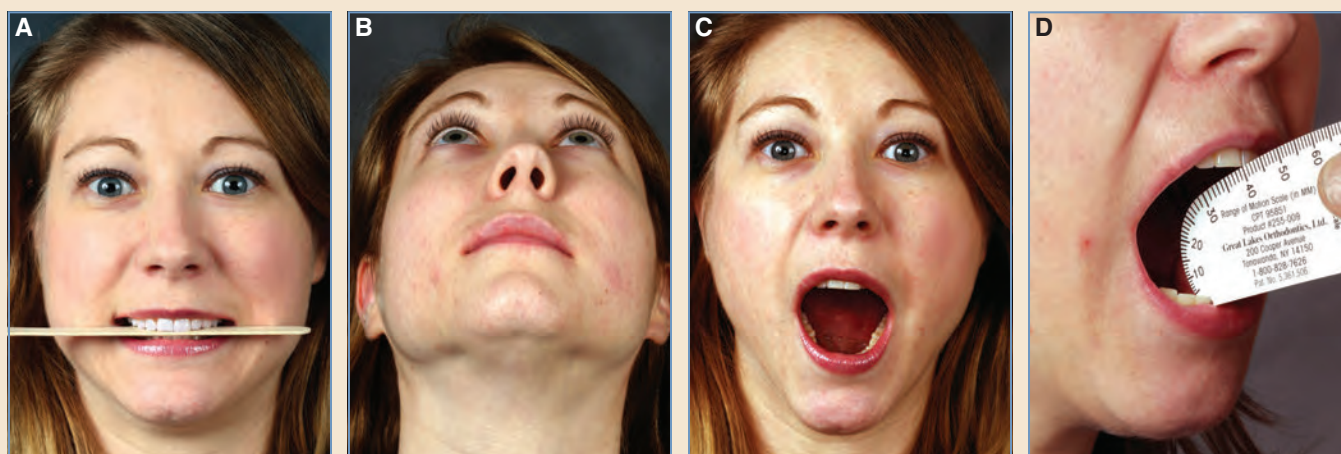
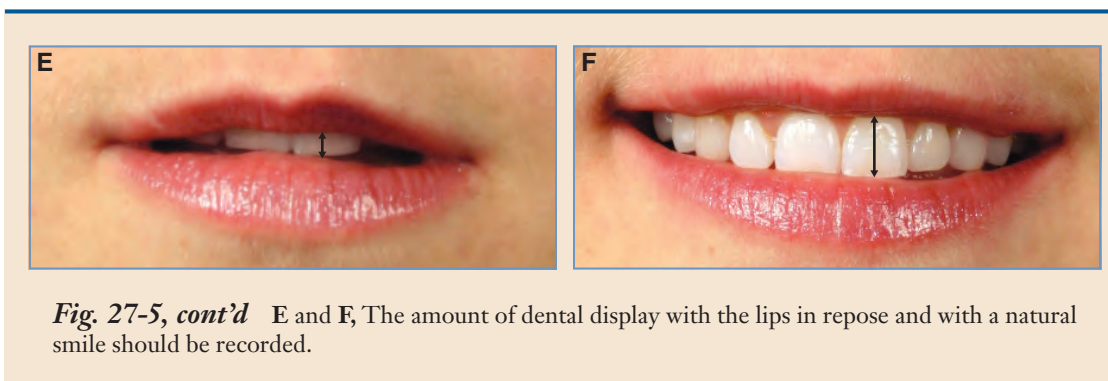


Fig. 27-5 A and B, The cant in the frontal plane and any rotational deformity in the transverse plane should be noted. C, With mouth opening, any deviation (lateral gnathism) should be noted. D, The maximal stomal opening from the maxillary incisal edge to mandibular incisal edge should be recorded.

Continued



will provide the most valuable information for determining the vertical and sagittal relationships of the face. In this view the clinical assessment includes a vertical height analysis, position of the midface relative to the cranial base (sagittal), the position of the mandible relative to the cranial base (sagittal), the position of the mandible relative to the maxilla (sagittal), the position of the chin (symphysis) relative to the mandible, and the position of the chin relative to the upper face, the columellar-lip angle, the depth of the labial mental sulcus, and the cervicomandibular angle. The oblique view adds the third dimension in assessing the degree of zygomatic projection and the perialar region. It is the most useful assessment for determining maxillary-malar deficiency, which may not be fully appreciated in the true lateral view.

The intraoral examination should focus on the overall state of oral hygiene, anatomic and functional tongue abnormalities, the presence or absence of teeth, dental crowding, tooth size discrepancies, the occlusal relationship (Angle classification, open bite, and crossbite; overjet and overbite relationships), the discrepancy between the occlusal relationship and centric occlusion, transverse width discrepancy between the maxillary and mandibular dental arches, and the degree of the curves of Spee and Wilson (Fig. 27-6).

The maxillary-mandibular occlusal relationship should be evaluated with the condyles seated within the glenoid fossa (centric relation) and the teeth lightly touching. When asked to show their bite, patients will frequently not necessarily be in centric relation. Instead, they may bite in their normal habitual occlusal relationship—that is, the occlusion these patients go into when they are unconsciously talking, eating, or swallowing. Many patients find it difficult to relax and allow the examiner to manipulate the mandible into the glenoid fossa (centric relation examination). An alternative is to ask the patient to place the tip of the tongue as far back on the roof of the mouth as possible while gently closing the teeth. Failure to appropriately determine the occlusal relationship in centric relation may lead to an improper diagnosis and a compromised outcome. In patients with dentofacial deformities, the centric relation, centric occlusion, and habitual occlusion are not necessarily equal.^{3,5}

Because temporomandibular joint disorders may coexist with dentofacial deformities, the masticatory function should be documented.^{15,16} The history should focus on headaches, muscle pain, clenching and grinding of teeth, limitations of opening, locking in the open or closed position, and clicking, popping, and pain with functioning of the temporomandibular joint. Patients should be questioned regarding the onset, duration, and factors that improve or worsen these symptoms. The muscles of mastication should be palpated and areas of tenderness noted. The timing of any clicking, grinding, or popping should be noted within the opening and closing cycles. The interincisal opening, protrusion, and lateral excursion should be recorded. The normal maximal stomal opening measures 40 to 56 mm, with lateral excursion in the range of 9 to 13 cm. The pattern of opening and closing with any lateral deviation should also be noted.

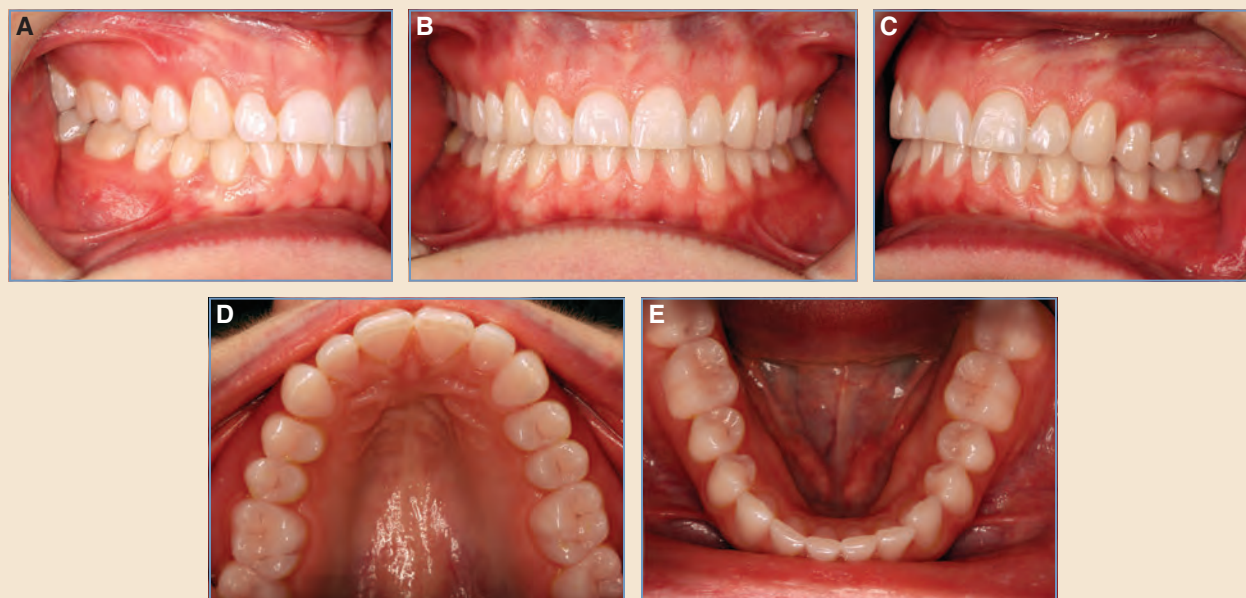


Fig. 27-6 A-C, The dentition in each arch should be assessed (oblique and frontal views). D and E, The occlusal relation between the arches (upper and lower) should be noted.

Based on the results of this clinical examination alone, a decision can be made in most circumstances regarding which jaw needs surgical repositioning to restore the normal anatomic relationship.

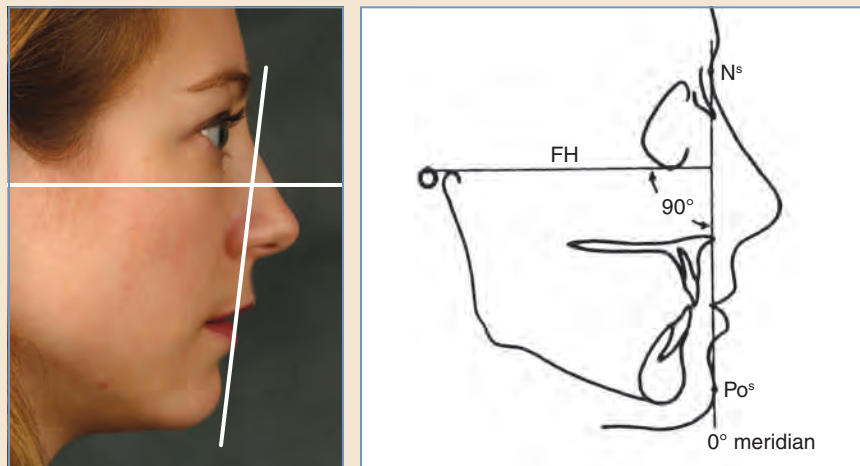
Photographic Documentation

Clinical photographs are essential to document the clinical evaluation and to allow photometric analysis.¹⁷ Soft tissue landmarks are used to obtain angular and linear measurements that can help quantitatively define what is “out of place.” Frontal, lateral, oblique, and intraoral photographs should be standardized.⁷ A clear acetate ruler can be placed next to the patient as the photographs are taken for reference measurement; otherwise, a proportionate type of analysis without relying on absolute values can be used. Facial balance is typically assessed by dividing the face into thirds. The upper third goes from the anterior hairline (trichion) to the glabella, the middle third is from the glabella to the subnasale, and the lower third is from the subnasale to the menton. When each of the thirds is equal, the face is said to be balanced and of “ideal” proportions. It is important to remember that facial proportions are only idealized concepts that have changed over time.¹⁸⁻²¹ Such rules merely provide a rule of thumb, and there is a wide range of aesthetically pleasing faces that defy such absolute canons (Figs. 27-7 and 27-8).

In recent years facial images have been captured by a variety of three-dimensional (3D) systems that have begun to transition from the research laboratory to the clinical setting. These provide the ability to “reproduce” the patient’s three-dimensional surface images of the face in a digital media that allows the clinician to manipulate the patient’s head as an object in virtual space and to make objective soft tissue cephalometric measurements. Although the expense of such systems is a significant limiting factor in routinely replacing the traditional two-dimensional (2D+ digital photography, it has the potential to become the standard in the future.

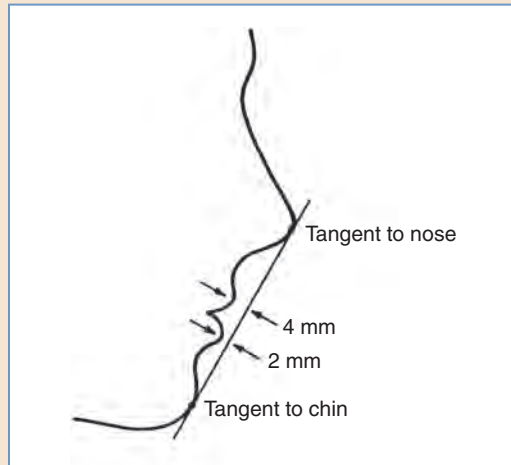


Fig. 27-7 Photometric analysis is used to assess facial symmetry, applying the rule of fifths and the rule of thirds.

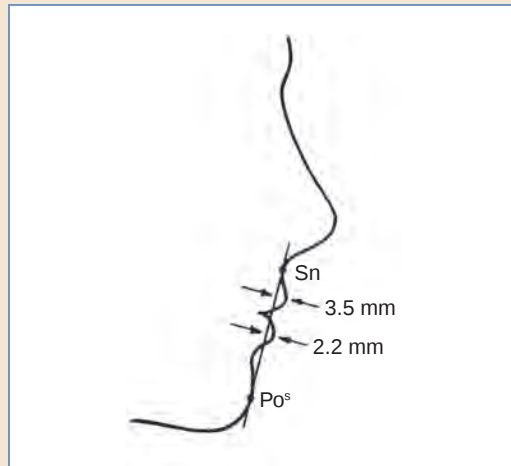


Gonzalez-Ulloa and Stevens

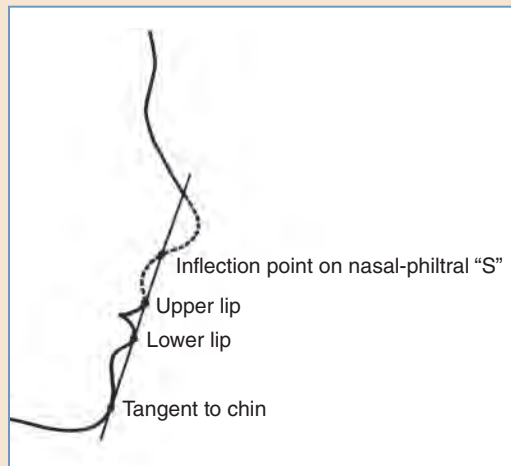
Fig. 27-8 Photometric analyses of the lower third of the face. (P_0 , Porion.)



Ricketts



Burstone



Steiner

Fig. 27-8, cont'd Photometric analyses of the lower third of the face. (*Po*, Porion.)

Skeletal Evaluation

Skeletal evaluation has traditionally relied on panorex and cephalometric radiographs.²² Panorex radiographs provide an overview of the stage of dental development, the mandibular anatomy, and any gross pathologic findings. Specific films such as occlusal and periapical views can be obtained to further assess the dentition, supporting bone, and interdental spaces. Cephalometric radiographs provide standardized skull/facial views that allow comparison over time to assess growth in individual patients and to compare individual patients against standardized population norms.²³⁻²⁵ Skeletal views can be obtained in lateral and frontal views. Lateral cephalometric radiographs allow an assessment of the elements of the dentofacial skeleton from a sagittal perspective. The maxilla can be related to the mandible, and each can be related to their position in the skull base. A soft tissue profile is simultaneously obtained, which facilitates relating the soft tissue to the facial skeleton. In addition, the upper and lower dentition can be related to each other and each to its own skeletal base, the maxilla, and the mandible.

Basic measurements, based on key anatomic landmarks, are obtained from the cephalometric radiographs and form the basis of cephalometric analysis, providing quantitative assessment (Fig. 27-9).

When cephalometric measurements are analyzed, no single measurement can summarize the dentofacial deformity. The measurements must be analyzed as “collections” or “groupings” to reflect the clinical picture. In addition, the analysis cannot rely on a single reference plane, the SN plane or the FH plane alone, but both must be used to correlate the spatial position of each of the jaws with stable structures. Numerous cephalometric analyses exist; each emphasizes particular skeletal and dental elements. Common analyses include those of Steiner, Ricketts, Delaire, Jarabak, and Sassouni, among others (Fig. 27-10).

Regardless of the analysis used, differences may occur in treatment planning that vary from the findings of the initial clinical assessment. In this instance, the surgeon should rely on the clinical examination. Although these analyses are invaluable, the treatment plan should not focus solely on correcting cephalometric abnormalities. The goal of treatment is not to normalize the patient's cephalometric measurements, but to make the facial appearance aesthetically pleasing while simultaneously establishing a functional occlusion.

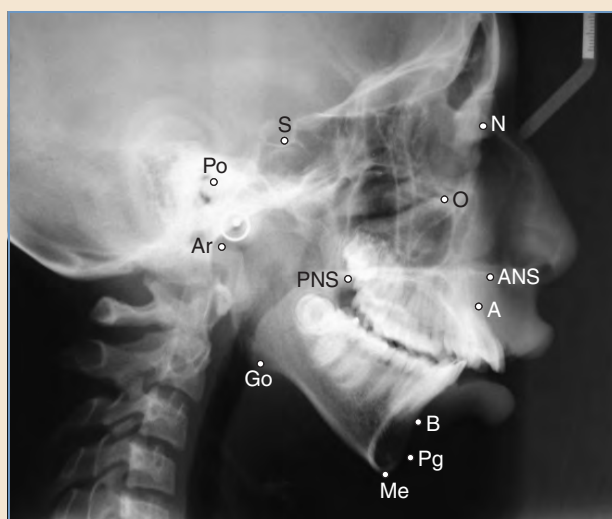


Fig. 27-9 The facial skeleton is analyzed using cephalometric radiographs with anatomic landmarks. (*A*, A point; *ANS*, anterior nasal spine; *Ar*, articulare; *B*, B point; *Go*, gonion; *Me*, menton; *N*, nasion; *O*, orbitale; *Pg*, pogonion; *PNS*, posterior nasal spine; *Po*, porion; *S*, Sella.)

Cephalometric analysis, when used appropriately within its limitations, can provide the objective evidence needed to guide but not determine treatment planning and assess treatment outcomes (Table 27-1).^{26,27} For the surgeon, the analysis must be clinically workable, simple to use, and directly correlated with the skeletal elements that can be repositioned.^{28,29} Most surgeons use a combination of analyses that incorporate Steiner's SNA, SNB, and ANB analysis for the

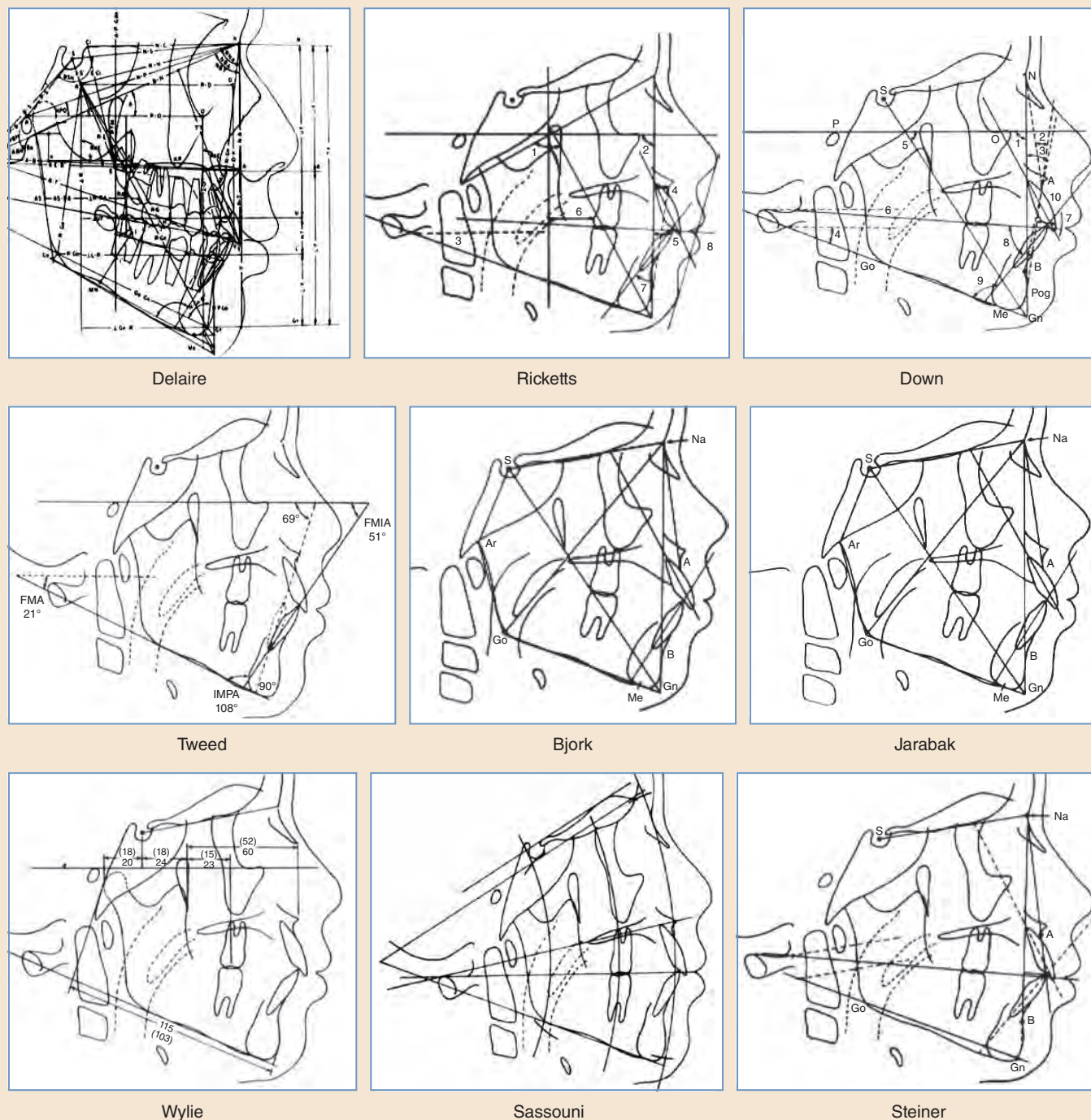


Fig. 27-10 Numerous cephalometric analyses exist to quantify the facial skeletal deformity. The choice of which analysis to use depends on the specific aspects of the deformity that interest the orthodontist and surgeon.

skeletal component (Fig. 27-11). However, comprehensive assessment requires the inclusion of not only skeletal but also soft tissue and dental components (Fig. 27-12).

Both photographic soft tissue and cephalometric skeletal analyses are based on static 2D images. Relying solely on these analyses has inherent limitations and may not correlate with surgical decisions that are based on clinical evaluation, which provides a 3D view and incorporates

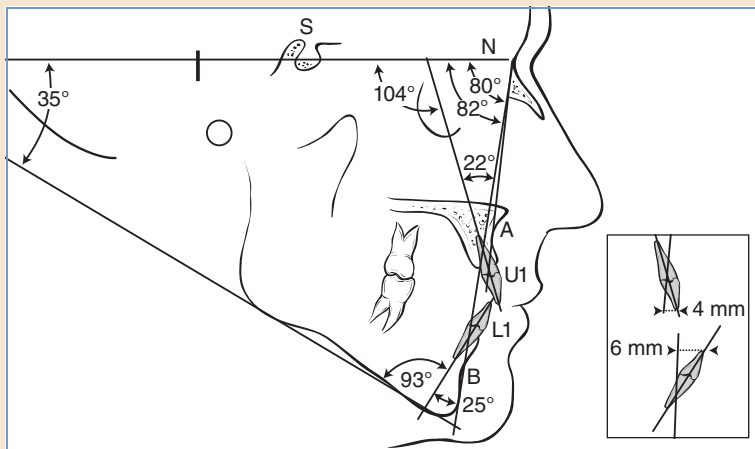


Fig. 27-11 Normative data in Steiner's Analysis (SNA, SNB, ANB). In craniofacial deformities, the SN length may be smaller than normal or the inclination of the cranial base may be steeper. This would affect the facial skeletal interpretation of the SNA, SNB, and ANB angles. If the S-N length is small, the SNA and SNB angles become less acute and would suggest a relative pseudoprognathism. As the S-N inclination increases, the SNA and SNB angles become more acute and suggest relative pseudoretrognathism.

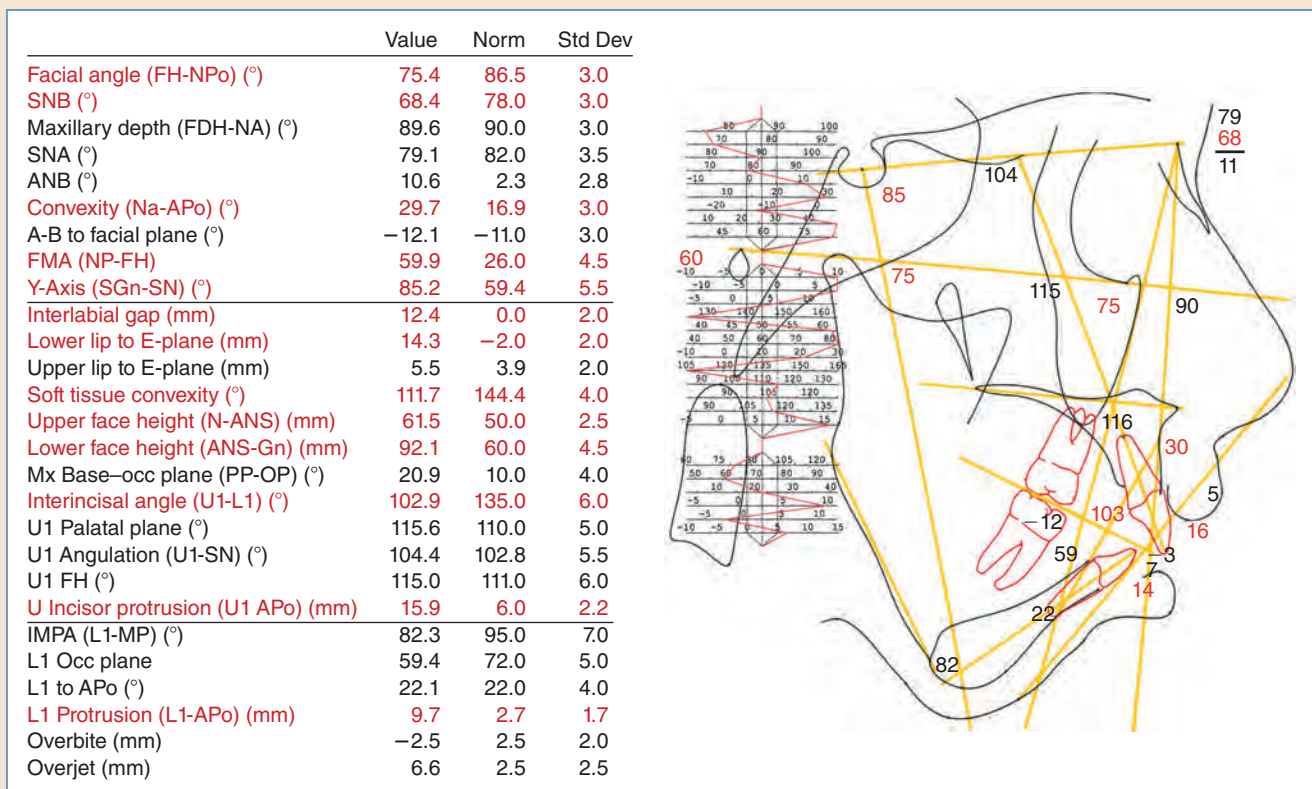


Fig. 27-12 Detailed cephalometric analysis that incorporates soft tissue, skeletal, and dental components.

dynamic hard and soft tissue skeletal relationships. Furthermore, cephalometric soft tissue and skeletal analysis attempts to quantify the dentofacial skeletal deformity in angular-linear measurements, which fails to take into account the complexity of the relationship between multidimensional surface contours of the soft tissue and the underlying skeletal framework. The final decision must be based on three-dimensional assessment of the deformity and not on normalizing the facial skeleton and soft tissue envelope to two-dimensional static normative data.

The emergence of low-dose radiation CT scanners, such as cone-beam computed tomography (CBCT) scanning, has increasingly replaced the use of traditional lateral and frontal cephalometric radiographs for the skeletal assessment of patients for orthognathic surgery. With a single scan, the three-dimensional facial skeletal structure can be captured and from which the

Table 27-1 Typical Presentation of Dentofacial Skeletal Deformities

Deformity	Clinical Features	Skeletal Assessment	Dental Assessment
Maxilla			
Sagittal deficiency	Concave facial profile Retrusive upper lip Acute nasolabial angle Alar base narrow Lack of dental display	SNA decreased SNB normal ANB decreased	Class III Maxillary dental crowding Maxillary incisors proclined Mandibular incisors normal or retroclined
Sagittal excess	Convex facial profile Obtuse nasolabial angle	SNA increased SNB normal ANB increased	
Vertical excess (long-face syndrome)	Convex profile Lower facial height increased Alar base constricted Nasolabial angle obtuse Excessive incisor show Excessive gingival show Lip incompetence Mentalis strain with lip closure Chin vertically long, retruded	Lower facial height increased SNA decreased SNB decreased ANB increased Mandibular plane angle steep Palatal-occlusal plane increased	Class II, class I Anterior open bite Maxillary arch constricted Curve of Spee flat accentuated Dental crowding
Vertical deficiency (short-face syndrome)	Concave facial profile Lower facial height decreased Nasolabial angle acute Alar base widened Lack of incisor show Edentulous appearance	Lower facial height decreased SNB increased ANB negative Palatal-occlusal plane decreased Mandibular plane angle acute	Class II, class I Deep bite Crowding Mandibular dentition Curve of Spee reversed
Mandible			
Deficiency	Convex profile Retruded chin Lower lip everted Deep labiomental crease Mentalis strain with lip closure	SNA normal SNB decreased ANB increased Ar-Gn decreased	Class II Mandibular incisors proclined Maxillary incisors retroclined Curve of Spee accentuated
Excess	Concave profile Midface appears deficient Lower third broad Lower lip thin	SNA normal SNB decreased ANB decreased	Class II Maxillary incisors proclined Mandibular incisors retroclined

traditional cephalometric radiographs and panorex can be digitally extracted. Moreover the ability to capture the facial skeleton in 3D allows assessment of the asymmetries in all three planes. The CBCT eliminates the inherent limitations of assessing the complexity of a three-dimensional facial skeleton with traditional 2D orthogonal radiographs (the lateral and frontal cephalometric radiographs). However, despite the detailed information useful to the surgeon provided by the CBCT, the use of conventional 2D lateral cephalometric and panorex radiographs remains the primary mode of skeletal assessment for the vast majority of the orthodontic practices because of the expense of the CBCT unit and the trade-off of increased radiation in routine cases. In the vast majority of referrals to the surgeon, the orthodontist will review the traditional 2D lateral cephalometric analysis with the surgeon, and the surgeon will determine the need to capture the three-dimensional facial skeleton with CBCT. Although a discussion of the pros and cons of CBCT versus conventional cephalometric radiographs is beyond the scope of this chapter, these are mentioned because advances in technology will continue to have an impact on clinical practice.

Occlusal Evaluation

Surgical movements of the maxilla and mandible inherently alter the maxillary-mandibular dental occlusion; as such, careful analysis of the dental arches is necessary, working with the orthodontist. The dental arches are studied with either traditional plaster/stone dental casts that can be hand manipulated to assess how well the dental arches are coordinated, or increasingly today, optical scanning is used, which allows digital manipulation of the dental arches on a computer. Assessment of the dental arches includes an analysis of space and arch length, transverse width discrepancies, position of the individual tooth within its own arch, and the relationship of the maxillary and mandibular dentition (Fig. 27-13). The stability of surgical repositioning of the maxilla and mandible will ultimately depend on achieving a stable occlusal relationship, and the use of dental casts will allow visualization of the role of supportive orthodontic management to achieve the skeletal surgical goals.^{3,5} The diagnosis is then established from a working list of problems generated from clinical and photographic evaluations, cephalometric analysis, and dental study models.

Thus, to correct dentofacial skeletal deformities, the role of the orthodontist is equal in importance to that of the surgeon, inasmuch as treatment requires the coordinated efforts of both. There must be a close working relationship between the orthodontist and surgeon, because the goals of presurgical orthodontic treatment are the opposite of the routine orthodontic treatment used to camouflage skeletal discrepancies in patients who choose not to pursue surgery or in situations in which the orthodontist and patient decide that the skeletal discrepancy is not significant enough to warrant surgical correction. Proper occlusion can be achieved with orthodontic treatment alone in minor skeletal discrepancies, and at times even in more significant discrepancies; however, it is frequently at the expense of facial appearance.

These same patients who have undergone orthodontic treatment with no consideration given to combined surgical-orthodontic management may seek plastic surgery procedures later in life to address residual skeletal deformities.³⁰ After proper occlusion is achieved, skeletal correction is compromised and frequently requires the use of prosthetic implants and soft tissue procedures to camouflage the deformity, the results of which are less than ideal.

The answer to the question of when is the problem too severe for orthodontic treatment alone depends on the severity and the patient's age at presentation. With the same degree of severity, orthodontic treatment in children who are still growing is likely to have a favorable influence and may help to avoid the need for orthognathic surgery, with the exception of clefts and craniofacial abnormalities. In adults with the same degree of severity, surgery is the only option. However, when the malocclusion persists in the growing child despite the use of orthodontic

treatment to modify growth, surgical correction should be considered either during active growth or when the patient is skeletally mature.

A successful outcome is one in which both the occlusal goals and aesthetic goals are achieved. In the opinion of many orthodontists, the limitations of orthodontic treatment lie within the confines of a positive overjet of +8 mm, a negative overjet of -4 mm, and a transverse width discrepancy of 3 mm.³¹ The well-known discrepancy diagram of Proffit and colleagues^{32,33} illustrates the envelopes of achievable outcomes based on orthodontic treatment alone (inner envelope), orthodontic treatment in a growing child (middle envelope), and with orthognathic surgery (outer envelope) (Fig. 27-14). The envelopes are asymmetrical and different for the maxilla and mandible. The precise dimensions of the envelopes are less important than the philosophic concepts that must be applied to each case.

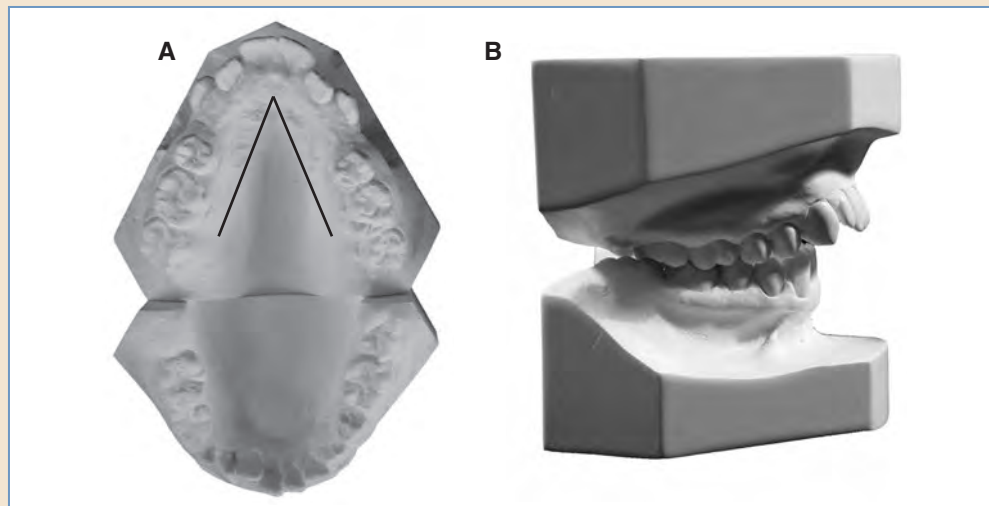


Fig. 27-13 Dental models are used to assess dental dysplasia. **A**, Arch form. **B**, Occlusion.

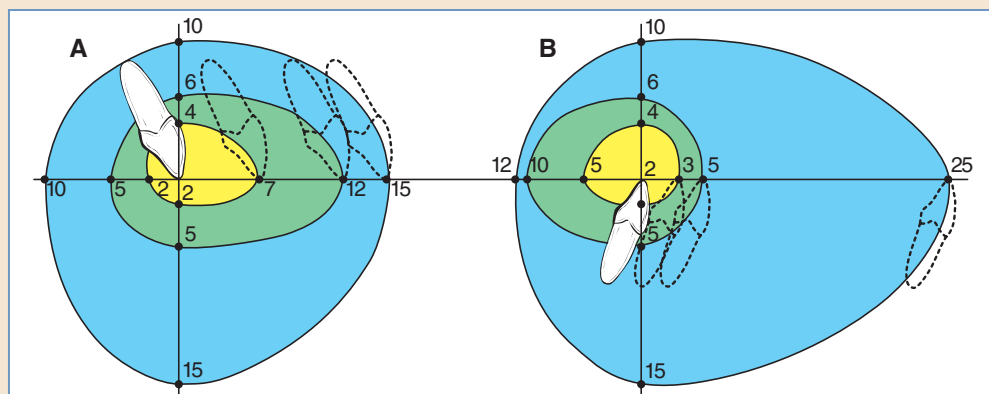


Fig. 27-14 The discrepancy diagram developed by Proffit's group for **A**, maxillary and **B**, mandibular dentition. The envelopes represent the amount of dental repositioning possible by orthodontic movement alone (yellow), orthodontic movement with growth modification (green), and with orthognathic surgery (blue). Note that the envelopes are not symmetrical and that, for example, orthodontic movement in the growing child is more effective with mandibular deficiency than with mandibular excess.

Growth and Development of the Maxillofacial Skeleton

A thorough knowledge of normal facial growth is important if abnormal patterns of growth are to be understood. Infants have a disproportionately large head and a small face, with eyes that appear large and widely set, a small nose, full cheeks, and a small lower jaw. It is this combination of features that makes infants so adorable. The same combination in skeletally mature adolescents would not elicit the same emotions. With the passage of time, the face emerges from beneath the cranium. The midface becomes more prominent, the eyes more deeply set beneath a brow, the nose more sharply defined, the lower jaw catches up, and the chin develops. Whether a person goes from being an adorable infant to an attractive or handsome adolescent depends on the proportions of the individual components. Thus a change in size and shape in all three dimensions occurs. The growth and development of the face from infancy to maturity occur at different rates, both spatially and temporally, where various elements of the face mature to differing extents and at different times (Fig. 27-15). Throughout this growth, the facial elements in their various stages of maturity remain interlocked anatomically and functionally.^{34,35}

By 3 or 4 years of age, the growth of the brain, and correspondingly the skull, slows considerably, and by approximately 6 years of age, the cranioorbital regions reach nearly 90% of the adult size.³⁶ In contrast to the deceleration in the cranial region, the facial bones begin to grow and develop throughout the remaining years of childhood and into adolescence. Whereas the midface (maxilla) reaches skeletal maturity in early adolescence, the lower facial skeleton (the mandible) continues to grow well into adolescence. There is a difference between the sexes; girls reach skeletal maturity in early adolescence (14 or 15 years of age) and boys in late adolescence and early adulthood (17 to 21 years of age). In addition, there are differences in growth in each of

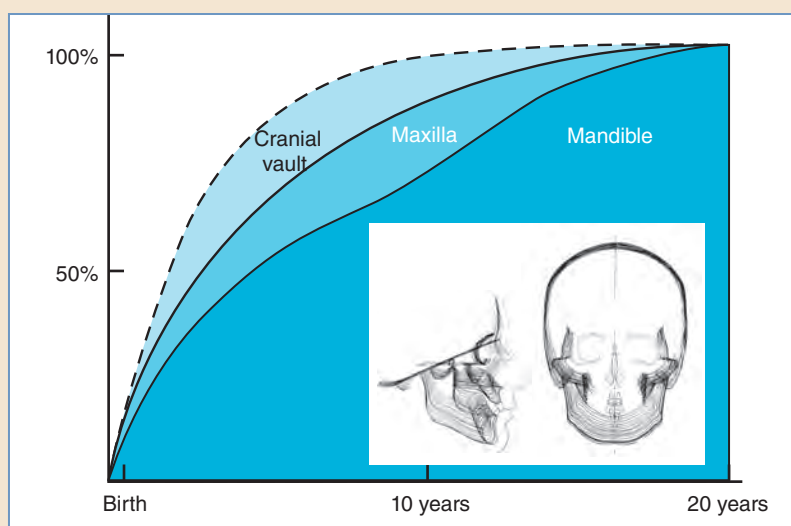


Fig. 27-15 Skeletal maturity of the cranial vault, maxilla, and mandible from infancy to skeletal maturity (100% of adult size). The cranial vault reaches maturity long before the midface, followed by the mandible.

the facial dimensions. The width of the face reaches skeletal maturity long before the final maturity in the vertical length of the face.³⁴⁻³⁷

The basicranium itself has a significant influence on facial form, as the posterior boundary of the facial mass coincides with the boundary of the anterior and middle cranial fossa. Early on, with expansile growth of the anterior and middle cranial fossa with brain growth, the nasomaxillary complex and mandible are affected. This results in anteroinferior displacement within the nasomaxillary complex and vertical displacement of the mandible. By 5 or 6 years of age, growth in the frontal lobe and anterior cranial fossa nearly ceases; however, growth of the middle cranial fossa continues for several more years, which adds to the forward displacement of the nasomaxillary complex and vertical displacement of the mandible.³⁸⁻⁴⁰

Simultaneously, within the facial mass itself, bone deposition occurs at the various facial sutures and surfaces to allow volumetric enlargement while undergoing resorption along opposing surfaces. Sagittal lengthening of the maxillary arch occurs, with progressive bone deposition along the posterior surface of the maxillary tuberosity. Simultaneously, bone deposition along the lateral surface at the tuberosity increases the transverse width of the arch. With this posterior growth, the entire maxilla undergoes forward displacement in an anterior direction. Similarly, with the malar complex, growth (bone deposition) occurs along the posterior and lateral surfaces while simultaneously the anterior and medial surfaces undergo resorption. The malar complex maintains its continuity with the maxilla and also undergoes a forward displacement with posterior growth.³⁸⁻⁴⁰

The body of the mandible, of necessity, must undergo lengthening to match the maxillary arch. The anterior surface of the ramus undergoes remodeling by converting ramus to corpus. To maintain the width of the ramus, bone is added posteriorly. This backward sagittal growth then results in forward displacement of the mandible as a whole, such as with the maxilla. At the posterior region of the ramus, there are two distinct areas of growth: the posterior edge of the ramus itself and the condyle. Although the posterior edge is a sagittal growth direction, the growth of the condyle results in an angular vector that is posterior and superior. The condylar growth results in the anteroinferior displacement of the mandible. The ramus becomes progressively more upright by the anterior remodeling that occurs at its base to lengthen the mandibular body, and simultaneously comparatively more bone deposition occurs at the base of the posterior edge of the ramus. Essentially this results in rotation of the ramus, and the condylar growth vector is correspondingly more vertical, with a decrease in the gonial angle. This vertical lengthening then continues long after the horizontal ramal width ceases. This allows simultaneous accommodation of the vertical growth of the nasomaxillary complex without changing the occlusal relationship. Any difference in this interlocking growth pattern between the maxilla and mandible will lead to a skeletal sagittal discrepancy and/or a rotational imbalance (Fig. 27-16). There must also be a corresponding match that occurs in the transverse width between the jaws.³⁸⁻⁴⁰

Dental development plays an important role in maxillary-mandibular alveolar development. There are two aspects. One is dental eruption itself, and the other is the “drifting” of the tooth as a whole within the alveolar process. Movement of the tooth itself involves a process of similar remodeling of the tooth, its socket, and the periodontal membrane. While growth is present, the orthodontist can guide the individual teeth into position, taking advantage of the active phase of development with braces. In contrast, other orthodontic interventions, such as face masks and restraining chin cups, can alter the vector of the displacements of the maxilla and mandible, including the dental unit as a whole by affecting the sutures and sites of regional growth. These orthodontic maneuvers can have a significant and stable impact on future facial skeletal development only while growth is active and can be modulated (orthodontic growth modification).⁴¹⁻⁴³

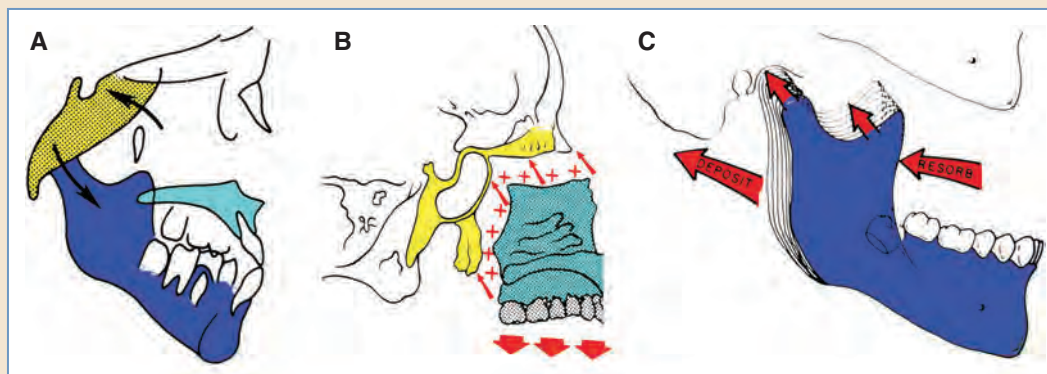


Fig. 27-16 Growth and development of the facial skeleton. **A**, Counterclockwise development of the basicranium (arrows) displaces the mandible anteriorly. **B**, With growth of the interface, the mid-face is displaced in the anteroinferior vector (arrow). **C**, With growth at the posterior ramal border and condyle, the mandible is displaced in the anteroinferior vector (arrow). These displacements of the maxilla and mandible must match to maintain the normal maxillary-mandibular occlusal relation. Any discrepancy leads to dentofacial skeletal deformity.

Surgical intervention at an early age is often not questioned in children who have a severe deformity that is a result of either a congenital or acquired condition. However, children who present with a moderate deformity with a dental malocclusion are the ones who have problems in the timing and sequencing of treatment. A fundamentally correct treatment plan instituted at the wrong time in a child's development can yield a poor result. Thus the timing of the intervention becomes the more critical issue facing the orthodontist and surgeon.^{44,45}

If the skeletal malalignment is minimal to moderate, it is possible to achieve correction by influencing the pattern of jaw growth with interceptive facial orthopedic treatment. This interceptive treatment must be initiated during the periods of active growth in early to late childhood but before the adolescent growth spurt. By the time the child has reached sexual maturity, it is too late to attempt such treatment.

It makes sense in most cases of moderate deformity to wait until skeletal maturity when planning for orthognathic surgery to avoid reoperation because of recurrence of the deformity with continued disproportionate growth. There is a variation between the sexes with regard to the timing of skeletal maturity. Serial cephalometric radiographs will objectively indicate deceleration of growth, and a hand-wrist radiograph can be used to determine skeletal age.⁴⁶⁻⁴⁸ However, there are circumstances in which relying solely on ensuring the stability of the correction to determine the timing of surgery may be inappropriate. Adolescents who have a poor self-image and those who are socially introverted or victims of ridicule can greatly benefit from early intervention. Similarly, functional indications—speech, airway, and occlusion—may dictate earlier intervention. The decision regarding when to operate is complex and requires that patients and their parents be well informed, frequently guided by input from other members of the team.

OVERVIEW OF ORTHODONTIC-SURGICAL MANAGEMENT

Treatment planning begins with a review of the various orthodontic and surgical options available that will address the list of problems. The primary decision that must be made is whether the deformity is significant enough to require surgical repositioning, or orthodontic alignment of the dentition alone can be achieved without significantly compromising facial aesthetics. If surgical intervention is contemplated, the question then arises regarding whether to intervene before the completion of skeletal growth or wait for skeletal maturity to eliminate the variability of subsequent growth and the need for further surgical intervention. After the decision is made to combine dental and surgical correction, the overall treatment must be carefully planned. Thus formulating a treatment plan requires close cooperation between the surgeon, dentist, orthodontist, and at times the restorative prosthodontist. Unlike many surgical procedures, the outcome depends not only on the surgical procedure itself but also on a multitude of factors that begin long before the actual surgery and control of the variables long after the surgery.⁴⁹ The management (Fig. 27-17) can be divided into the following five phases: (1) the preorthodontic preparatory phase (dental care and oral hygiene), (2) presurgical orthodontic treatment phase, (3) surgical phase, (4) postsurgical orthodontic phase, and (5) prosthodontic and restorative treatment phase.

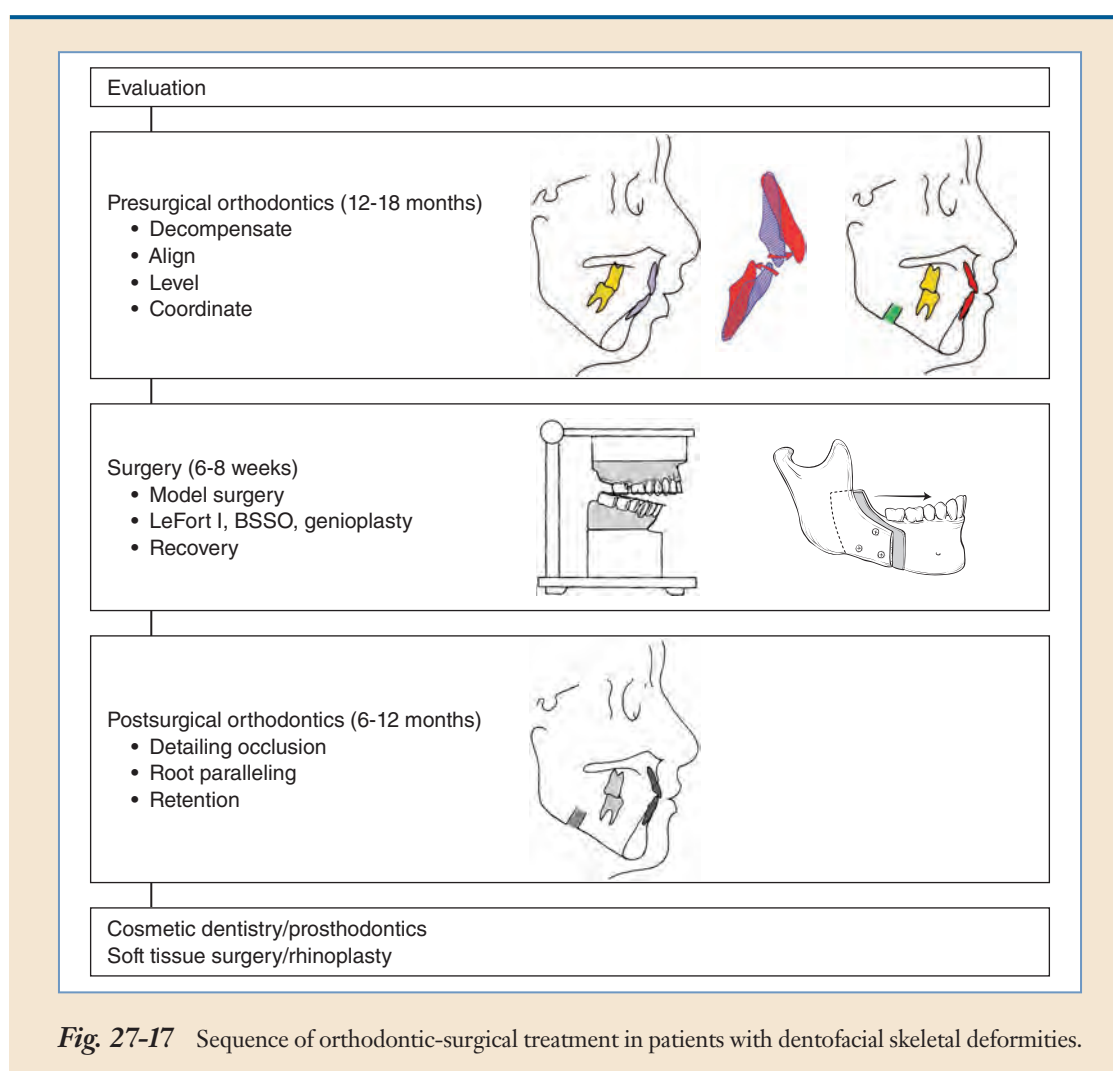


Fig. 27-17 Sequence of orthodontic-surgical treatment in patients with dentofacial skeletal deformities.

Dental Care and Oral Hygiene

Before the formal start of the presurgical orthodontic treatment phase, acute periodontal disease and caries must be managed as part of this initial phase.⁵⁰⁻⁵² Patients with periodontal disease and gingivitis have an increased risk of exacerbation with long-term application of orthodontic appliances, tooth movement, and during the postoperative period. Gingival inflammation must be reduced with cleaning and good oral hygiene. Poorly attached gingiva must be treated before any orthodontic tooth movement. Decisions must be made regarding whether carious dentition can be either temporarily or permanently restored, whether any endodontic treatment is required, or whether teeth should be extracted.

Presurgical Orthodontic Treatment

Once the state of oral hygiene is maximized, the presurgical orthodontic treatment phase can begin.⁵³⁻⁵⁷ With skeletal discrepancies, the teeth naturally compensate to establish as functional an occlusion as possible within the limitations of the deformity. The goal of the presurgical orthodontic preparation is to remove the dental compensations to reveal the true extent of the skeletal deformity. The teeth are orthodontically repositioned to their ideal position (axial inclination and alignment) within their respective maxillary and mandibular dental arches. Thus the occlusion is made worse until surgical correction occurs. Without appropriate dental decompensation, the surgeon is limited by the tooth position to fully correct the skeletal deformity. Thus the outcome depends on both the surgeon and orthodontist. The decompensation must be accomplished in all three dimensions: the sagittal plane (anteroposterior), the coronal plane (vertical), and the transverse plane (width) (Fig. 27-18).

In the transverse plane, the maxillary and mandibular dental arches need to be coordinated with the postsurgical occlusion (Fig. 27-19). Because of the relative transverse arch width discrepancy with class II and class III malocclusions, the dentoalveolar compensations must be removed. As with removal of the sagittal compensations, the arch width discrepancy is worsened. Depending on the discrepancy, the options are transpalatal orthodontic expansion with or without surgical assistance versus segmental maxillary osteotomies at the time of the formal orthognathic surgical procedure. Expansion may be accomplished orthodontically alone with a palatal expansion device if the midpalatal suture is open; otherwise, correction may require surgical assistance with an osteotomy. When interdental osteotomies are needed, orthodontic root divergence is necessary. Minor width discrepancies may be acceptable with some degree of orthodontic compensation and can frequently be corrected after surgery.

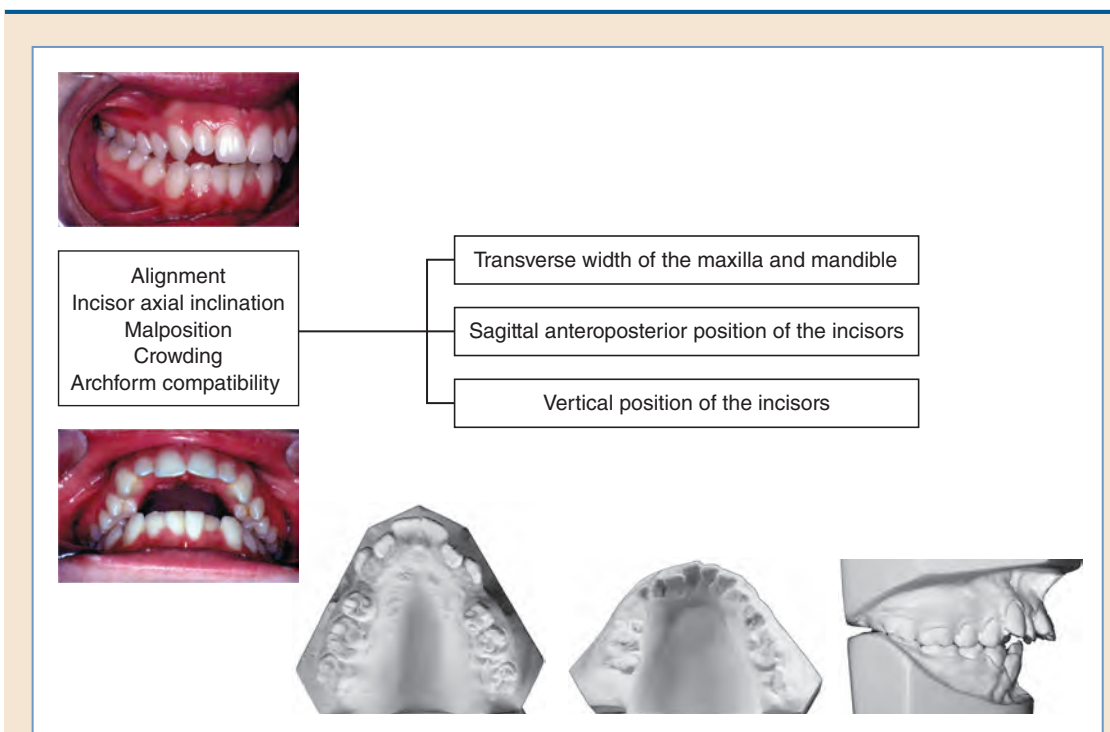


Fig. 27-18 Overview of presurgical orthodontic management.

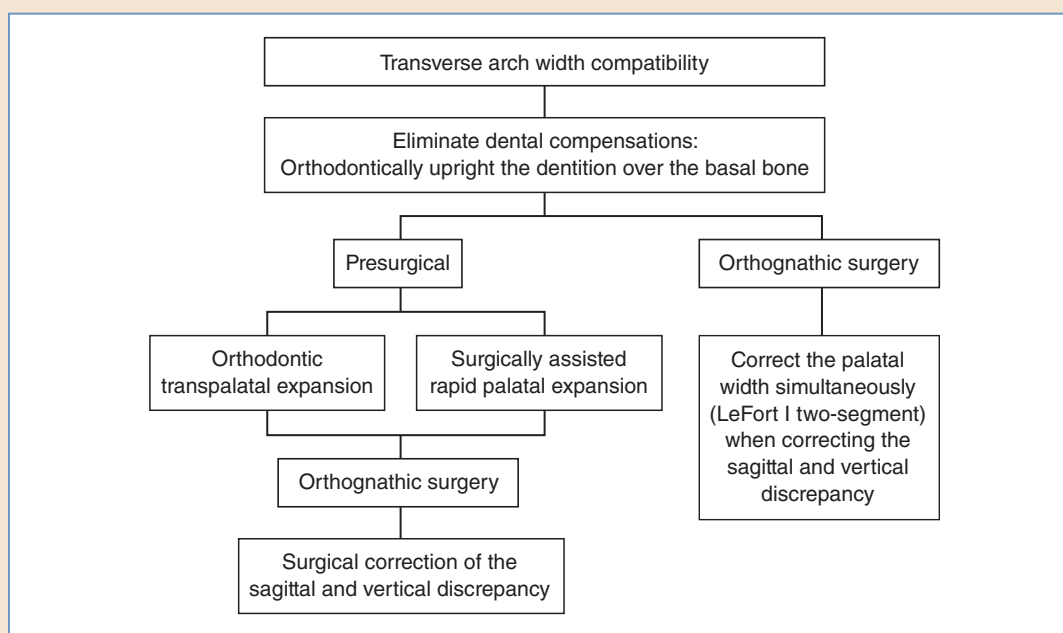


Fig. 27-19 Orthodontic management of the transverse component of dentofacial skeletal deformities.

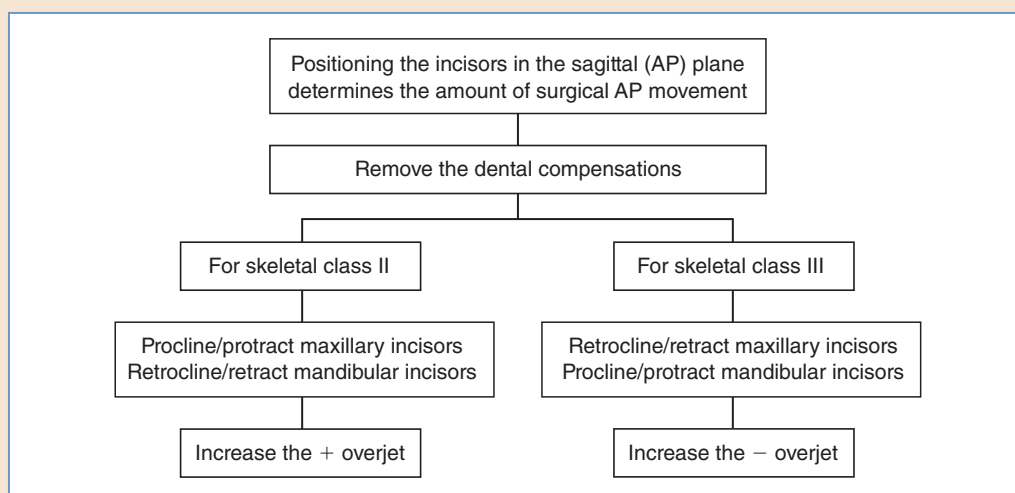


Fig. 27-20 Orthodontic management of the sagittal (anteroposterior [AP]) component of dentofacial skeletal deformities.

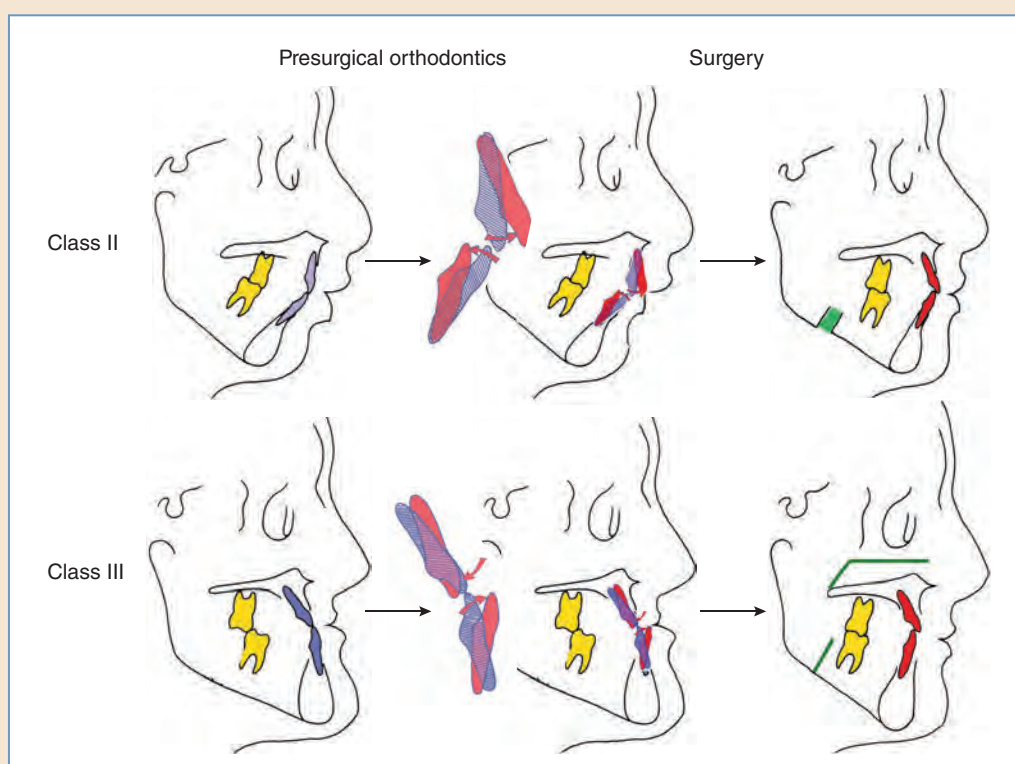
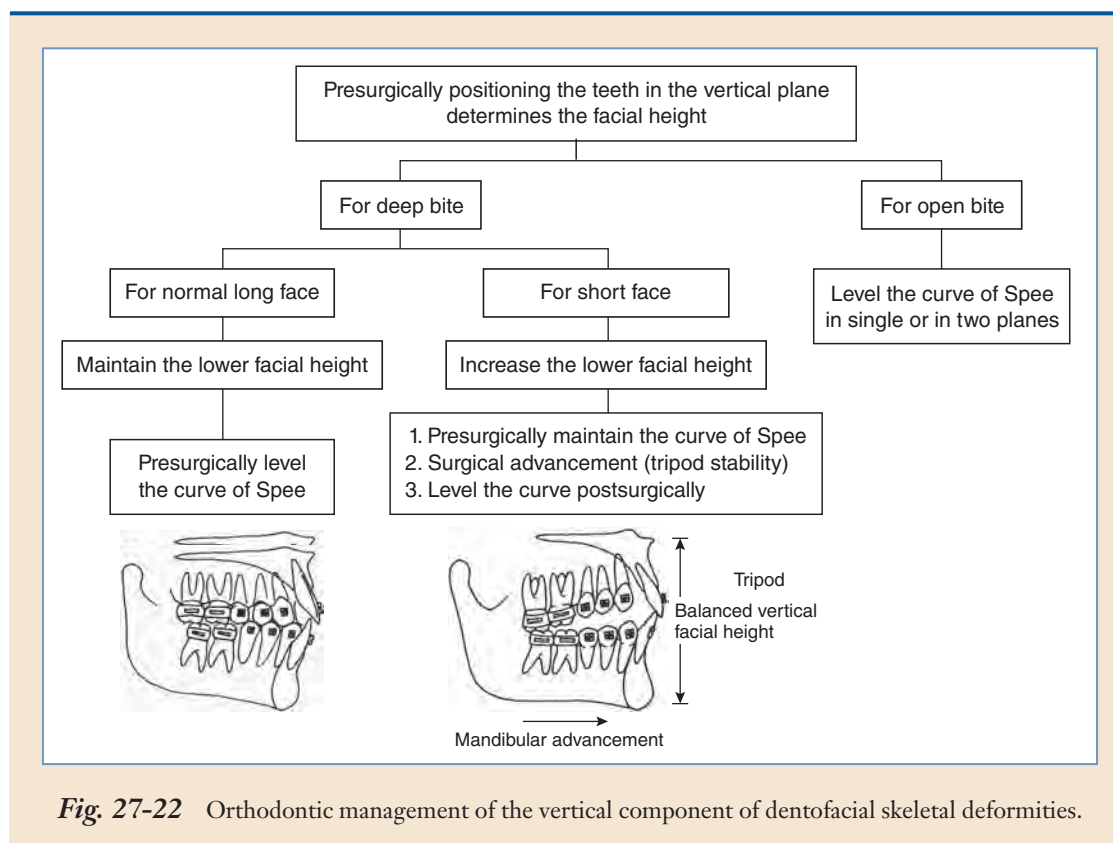


Fig. 27-21 Presurgical dental decompensation in the sagittal plane for class II and class III malocclusion. A goal of presurgical orthodontic management is to maximally decompensate the anterior dentition from pretreatment (*blue*) to presurgical (*red*) incisor position. As a consequence, the occlusion becomes worse. This allows the surgeon to reposition the skeleton so that when surgery is complete, the incisors are in an appropriate position with respect to their dental bases.

Anteroposterior decompensation involves correction of the maxillary and mandibular incisors (Figs. 27-20 and 27-21). In the typical class II malocclusion, the maxillary incisors are retroclined, and the mandibular incisors are proclined to allow a functional bite as a natural compensation to the skeletal deformity. Thus the decompensation involves orthodontically uprighting the proclined mandibular incisors. This may require removal of the mandibular first molars to relieve crowding if there is insufficient arch length to accommodate retraction and alignment of the incisors. In the maxilla, the opposite occurs. Decompensation of the maxillary incisors by proclination relieves any crowding typically without the need for extraction. Decompensation of class III malocclusion occurs in the reverse pattern.

In the vertical plane, dental decompensation of the anterior dentition depends on two factors: the amount of dental display of the maxillary incisors in relation to the upper lip and the lower anterior facial height (Fig. 27-22). When there is excessive dental display with gingival show in skeletal anterior open bite deformities, the extruded maxillary teeth that compensate for the open bite can be intruded orthodontically to a limited extent to level it in a single occlusal plane, with maxillary skeletal impaction surgery accomplishing most of the correction. Orthodontic correction alone will yield an unstable result. Frequently the posterior teeth may be in a significantly different occlusal plane from the anterior dentition. In such cases, if segmental maxillary repositioning is needed, the presurgical orthodontic treatment correspondingly involves segmental leveling in two different planes. In such cases, root diversion is needed to allow interdental osteotomies for a multisegmented LeFort I procedure.

In patients with a deep anterior incisor overbite, the decision of whether to level the occlusal plane depends on the lower facial height. When the lower facial height is normal, the deep bite is corrected by presurgically intruding the incisors. In contrast, when the lower facial height is



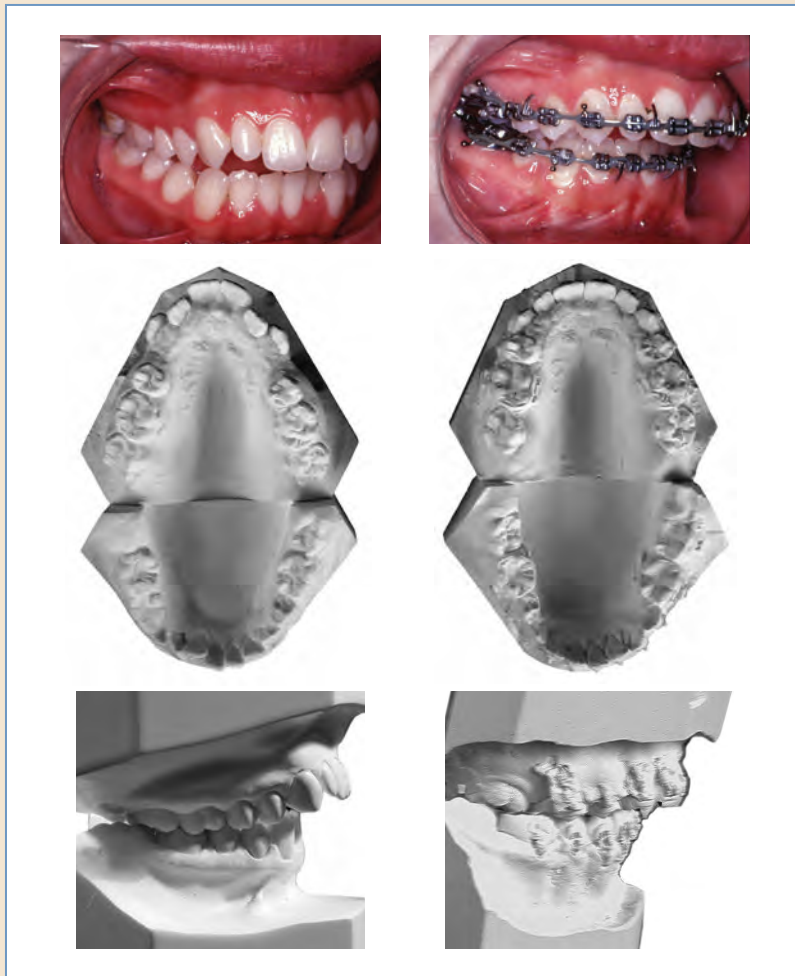


Fig. 27-23 Presurgical orthodontic management. The dentition has been aligned within the dental arches and decompensated to allow skeletal correction.

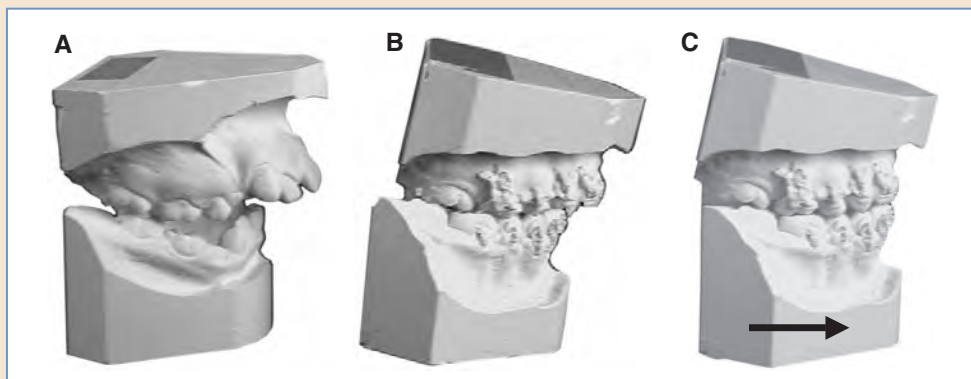


Fig. 27-24 Presurgical orthodontic management. **A**, Pretreatment. **B**, Orthodontic treatment without advancement. **C**, The maxillary-mandibular arches are well coordinated for surgery to ensure stability.

reduced, as in deep class II skeletal conditions, the arches are not leveled presurgically but instead after mandibular advancement. By maintaining the curve of Spee, the mandibular advancement will result in a “tripod” occlusion, with the incisors anteriorly and the molars posteriorly on either side. This will allow a natural increase in the lower facial height. The resulting lateral open bite will then be closed by extruding the mandibular premolars and leveling the curve of Spee. If the occlusal plane was instead leveled presurgically, correction of the lower facial height will then require osseous genioplasty.

In the transverse plane, the maxillary and mandibular dental arches need to be coordinated with the postsurgical occlusion. Because of the relative transverse arch width discrepancy with class II and class III malocclusions, the dentoalveolar compensations must be removed. As with removal of the sagittal compensations, the arch width discrepancy is worsened. Depending on the discrepancy, the options are transpalatal orthodontic expansion with or without surgical assistance versus segmental maxillary osteotomies at the time of the formal orthognathic surgical procedure. Expansion may be accomplished orthodontically alone with a palatal expansion device if the midpalatal suture is open; otherwise, this may require surgical assistance with an osteotomy. When interdental osteotomies are needed, orthodontic root divergence is necessary. Minor width discrepancies may be acceptable with some degree of orthodontic compensation and can frequently be corrected postoperatively.

If a mandibular osteotomy, such as sagittal split, is contemplated, it is generally wise to have the mandibular third molars extracted well in advance (typically 8 to 12 months) to minimize the risk of an unfavorable osteotomy and to allow internal fixation.⁵⁸ During this phase, the dentition is orthodontically aligned within the respective dental arches, the curve of Spee is leveled, and the anterior dentition is decompensated. This is done independently within each of the dental arches with no attempt to correct the occlusion. Because of this, the malocclusion is frequently made worse, and patients must be made aware of this. However, the maxillary and mandibular dentition are coordinated with each other as if the surgery had been accomplished with the use of dental casts, which can be freely manipulated. Progressive dental models are obtained throughout the orthodontic treatment, and when they can be hand coordinated with stable occlusion, then planning for surgery can begin (Figs. 27-23 and 27-24). If segmental osteotomies are planned, the dentition must be prepared accordingly. If osteotomies are not through planned extraction sites, interdental space must then be orthodontically prepared with the roots widely divergent to prevent injury, with the space favorably closed at the time of surgery. Periapical and panorex films should be used to evaluate the space for osteotomies. Segmental dental casts will confirm the readiness for surgery.

The orthodontist will then typically replace the archwire with a heavy rectangular wire ligated to the orthodontic brackets and surgical hooks to permit intraoperative and postoperative maxillary-mandibular fixation. At the time of surgery, there should be no active dental movement, and the orthodontic wire should remain passive. If segmental osteotomies are planned, the archwire should likewise be segmented before surgery. This presurgical phase typically varies from 6 to 18 months, depending on what needs to be accomplished to maximize the final surgical stability at the occlusal level.

Surgical Phase

The surgical phase consists of three components: surgical planning, the operation, and the immediate recovery phase.

Surgical Planning

The surgical phase begins approximately 4 weeks before the surgical date. This starts with a clinical reevaluation of the patient and obtaining presurgical planning records.^{27,59,60} The clinical

focus should include reconfirming with the patient whether a maxillary, mandibular, or double jaw procedure is to be performed. Dental midlines in relation to each other and in relation to the facial midline should be carefully recorded. Orthodontic movement of dentition alone will subtly alter facial features such as lip posture and the amount of dental display, and such changes need to be taken into account when planning the final skeletal movement. Key points should be discussed with the patient, such as the amount of dental display, appearance of the smile, changes in the nasal-columellar contour, alar base and nasal tip position, chin position, and any residual asymmetry that inevitably becomes more noticeable by the patient after the surgery.

If only a single-jaw procedure is planned, conventional cephalometric analysis and an acrylic splint fabrication from presurgical dental casts mounted on a simple hinge or Galetti-type articulator are sufficient to execute the surgical procedure (Fig. 27-25). The splint is needed to intra-operatively index the relative position of the maxillary dental arch to the mandibular dental arch. For upper jaw surgery only, the vertical position of the maxilla is primarily determined by the desired dental display and the appearance of the smile. With lower surgery alone, the mandible

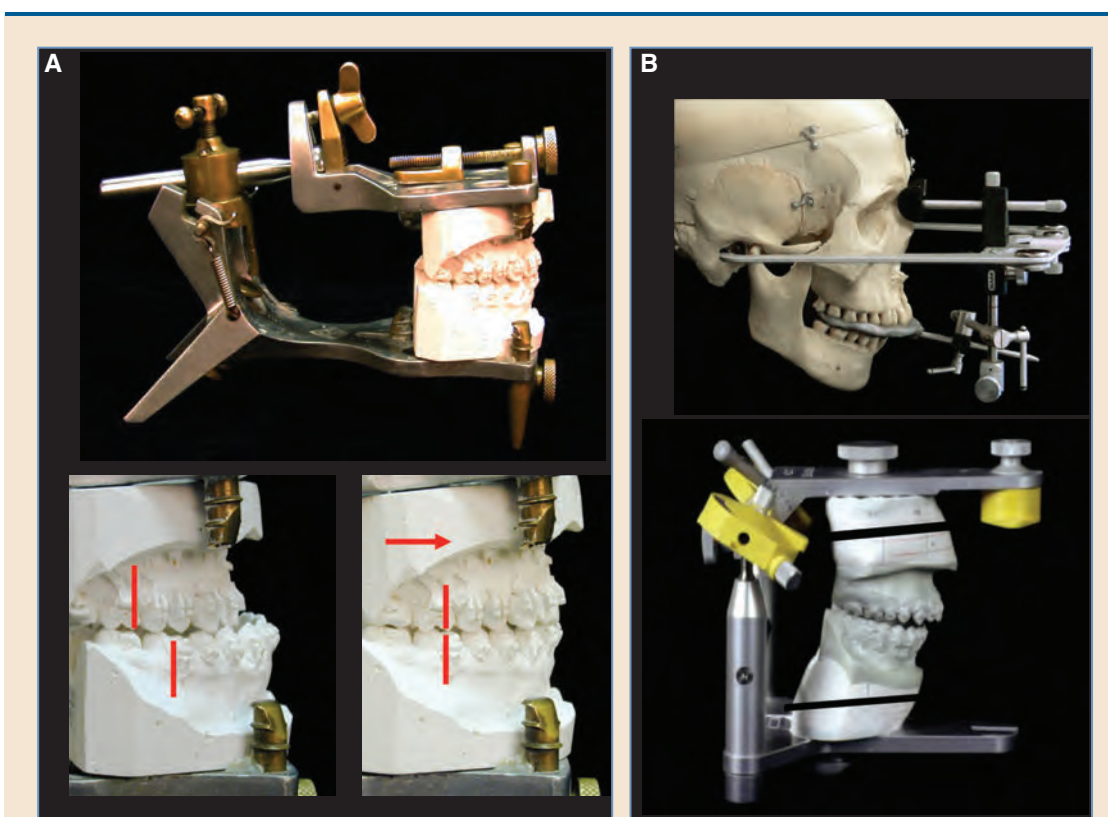


Fig. 27-25 A, Model surgery and splint fabrication for a single-jaw procedure with a hinge-type articulator (Galetti). This is independent of the skull base, because only the relative position is needed of the jaw that is being operated on to the jaw that is not. B, Facebow transfer and model surgery for a two-jaw procedure in which the relation of the jaws to the skull base (Frankfort horizontal) is needed to appropriately position the maxilla and mandible.

is positioned spatially by the fixed maxillary dental arch. Routine conventional 2D cephalometric analysis of the initial position and the predicted position is sufficient to guide the surgeon.^{26,61-65}

However, currently, with the exception of the simplest of orthognathic cases (single jaw with relative facial symmetry), we develop a composite 3D virtual model of the patient by fusing the 3D data sets of the CBCT scan of the facial skeleton with the digitally scanned dental models and the 3D surface color and texture images of the patient (3D photos). The composite model is developed in the digital environment with one of a number of virtual surgical planning (VSP) software systems that are now commercially available. The software allows the surgeon to simulate the various surgical procedures by tailoring the osteotomies and repositioning the various skeletal elements to the patient's specific deformity.

The ability to visualize the surgical simulation in 3D allows the surgeon to consider the asymmetry in the three orthogonal planes and to visualize osteotomy gaps and bone interferences that are likely to become evident during the operative procedure. Currently for us, 3D VSP has replaced the traditional approach of using facebow mounted dental casts and model surgery because of the inherent inaccuracies and limitations when applying it to patients with congenital deformities and facial asymmetries that require two-jaw surgery. Moreover, the “model” surgery in the virtual environment of the digital computer using the patient's facial skeleton is a more realistic and more relevant simulation for the surgeon⁶⁶⁻⁷¹ (Fig. 27-26).

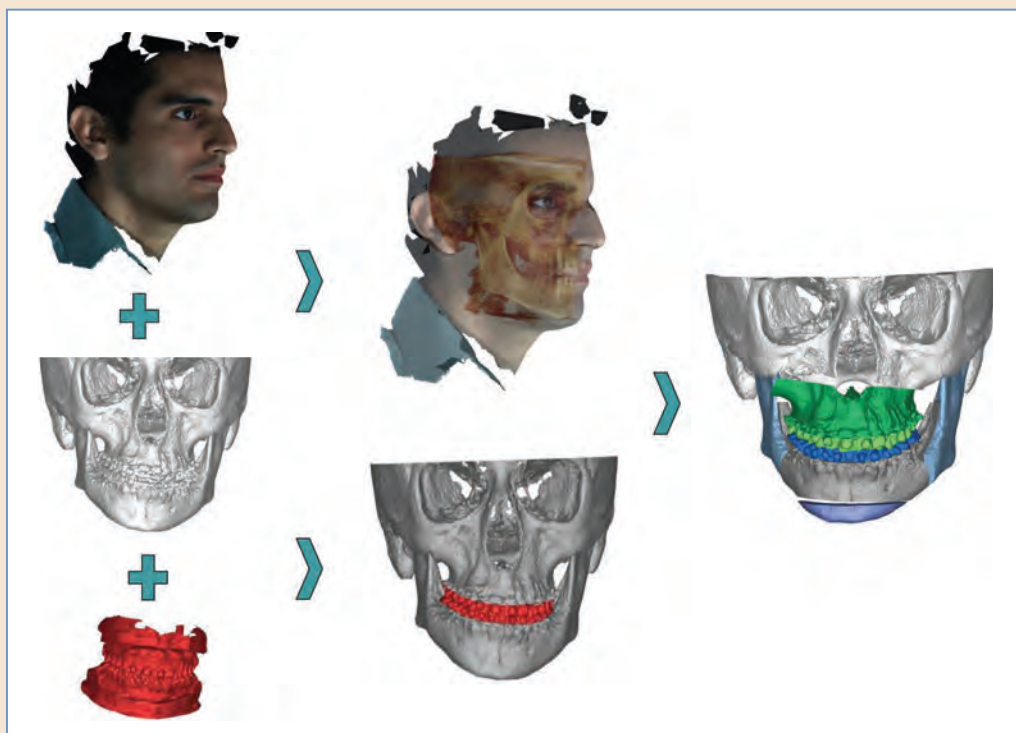
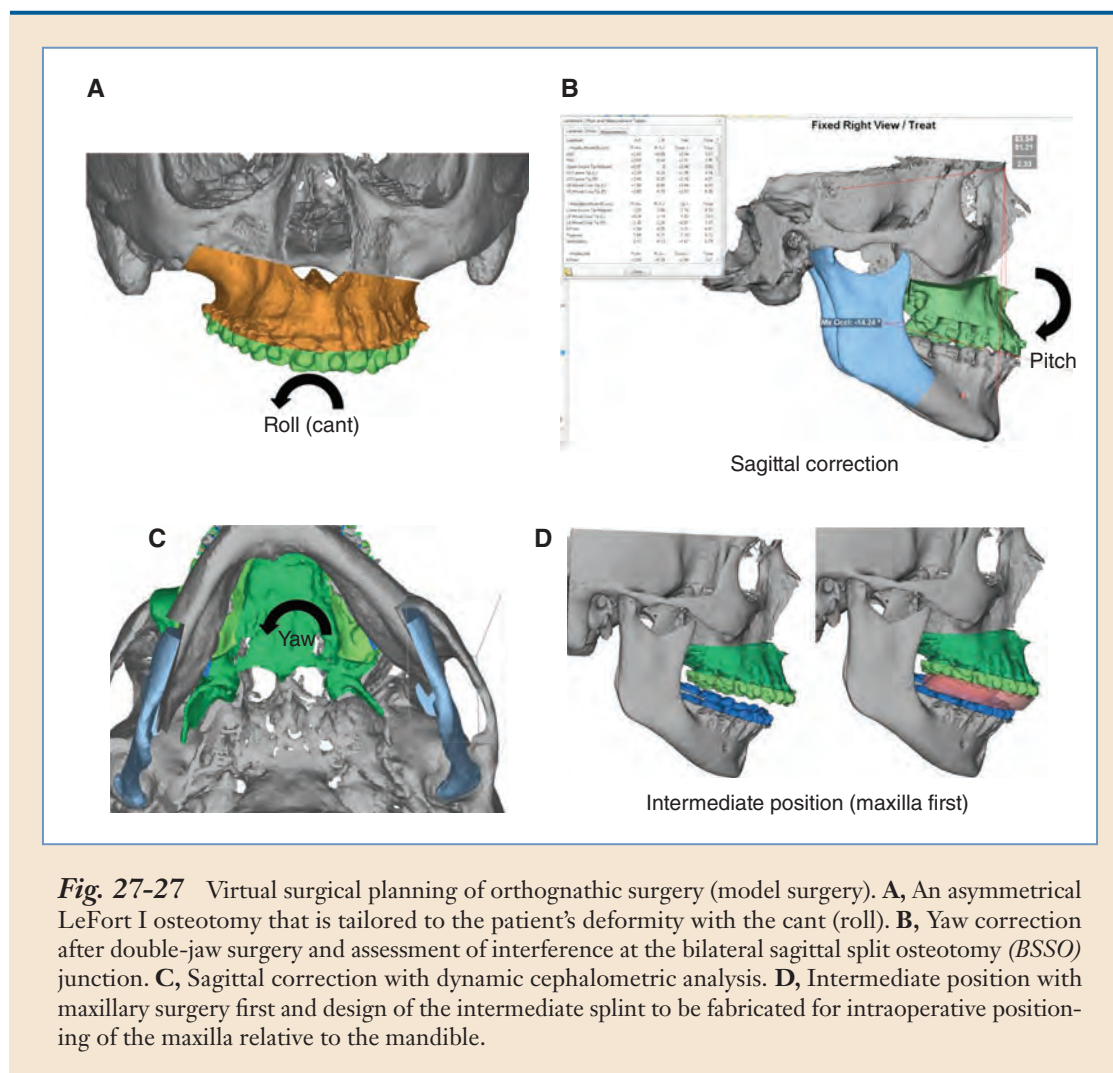


Fig. 27-26 VSP (model surgery) is based on the use of a composite model of the patient that “fuses” the 3D data sets of the CBCT scan, the dental models, and 3D photographic images. This replaces the model surgery using facebow mounted plaster/stone dental casts shown in Fig. 27-25, B.



A practical approach to VSP is to first correct the facial asymmetry in the frontal plane. This will then convert a complex case of asymmetry into a more straightforward and familiar orthognathic procedure that focuses on correcting the sagittal (lateral) discrepancy only (Fig. 27-27).

Even though the CBCT provides a 3D view of the facial skeleton, for analysis and correction of the dentofacial deformity, it is best to begin initially from the lateral view, with the standard lateral cephalometric view extracted from the CBCT. Thus our typical two-jaw sequence in simulation using VSP software is outlined in Table 27-2.

Steps 5, 6, and 7 establish frontal symmetry. Step 8 is the conventional and more familiar orthognathic 2D planning and sagittal (profile) correction. With the maxilla in the optimal position, steps 10, 11, and 12 reposition the mandible to the maxilla. Step 13 may require repeating steps 7 through 12. Correcting the yaw of the maxilla and mandible can be difficult. Step 15 repositions the chin to the desired position.

Table 27-2 Steps in 3D Computer-Assisted Virtual Surgical Planning of Orthognathic Surgery

Steps		Environment
1	Collect data from clinical assessment: 2D and 3D photographs, CBCT, dental models	Surgical records
2	Create virtual composite model of the patient	Virtual environment
3	Assign skeletal and soft tissue landmarks for cephalometric analysis in 3D and 2D	VSP software
4	Perform LeFort I osteotomy tailored to the patient's skeletal deformity	
5	Correct the maxillary asymmetry in the frontal plane "roll"	
6	Correct the maxillary dental midline to the facial skeletal midline "translation"	
7	Correct the maxillary asymmetry in the transverse plane "yaw"	
8	Reposition the maxilla in the sagittal plane to the desired position (dental display, smile, occlusal plane, incisor angulation); 2D cephalometrics	
9	Design an intermediate surgical splint based on the simulation of the upper jaw surgery first	Physical environment CAD-CAM 3D fabrication
10	Perform BSSO of the mandible	Virtual environment
11	Reposition the mandible to coordinate with the final desired occlusion	VSP software
12	Reposition the proximal ramal components to a more appropriate position relative to the mandibular body	
13	Assess, and if needed, correct the mandibular yaw; this may require repeating the sequence of steps 7-12	
14	Design the final surgical splint based on the lower jaw surgery second	Physical environment CAD-CAM 3D fabrication
15	Perform osseous genioplasty	Virtual environment VSP software

When repositioning of both jaws is planned, an intermediate splint (step 7), in addition to the final splint (step 11), is fabricated with 3D digital printing. These replace the traditional acrylic splint laboratory fabrication with facebow mounted plaster/stone model surgery. These splints then allow accurate intraoperative positioning of the maxilla and mandibular dentoalveolar segments. In addition, the surgical splints are important to ensure postoperative stability when the maxillary and mandibular arches are insufficiently coordinated for maximal intercuspal interdigitation.

Surgery

Surgery is then executed with the use of surgical splints to guide the intraoperative repositioning of the maxillary and mandibular segments. The surgical steps follow that of the VSP simulation. The details of the surgical technique are described later in a separate section.

Postoperative Recovery

In the immediate postoperative period, airway management is of primary concern, and patients require close monitoring. Most patients are discharged from the hospital after an overnight stay for hemodynamic and airway monitoring. The discharge instructions that are reviewed with the patient and family focus on controlling pain or discomfort primarily with the use of nonsteroidal antiinflammatory agents, supplemented by the occasional use of narcotic medications, cold compresses for edema and comfort, maintaining oral and nasal hygiene, a diet ranging from pureed to mechanically soft foods with maintenance of calories, proteins, and vitamins, and the importance of early ambulation and a return to normal daily activities.

A panorex/cephalometric radiograph in the early postoperative period is obtained to ensure that the condyles are in their appropriate positions. If the condyles are significantly displaced with a postoperative occlusion not anticipated, the patient will likely need to return to surgery and osteosynthesis will be redone.

Postoperative surgical practices vary, depending on the surgeon's preferences and the stability that the surgeon believes is obtained at the time of surgery. This has varied from maintenance of maxillary-mandibular fixation with wire to using only guiding dental elastics for varying periods, typically 2 to 6 weeks. Within this period the surgical splint, if used, is removed, and patients are instructed regarding how to increase the maxillary-mandibular range of motion that has been limited until now and to gradually return to their usual diet. Early function of the temporomandibular joint with continual assessment of the occlusion will allow the surgeon and orthodontist to use dental elastics to guide the occlusion, because internal fixation is "semirigid." Patients are followed closely, and if there is any change in occlusion, elastics can be applied accordingly in the appropriate vector to guide the occlusion until bony healing is complete.

Our experience has been that most patients recover by 2 weeks after surgery, and by the third week have returned to school and work, with appropriate limitations.

Postsurgical Orthodontic Management

The postsurgical orthodontic phase typically begins 4 to 8 weeks after surgery. Patients then return to the orthodontist for completion of the dental alignment with the relative position of the skeletal bases in their final position. Any remaining interdental spaces are closed, and the dentition is brought into an optimal intercusp relationship. This phase typically lasts for approximately 6 to 12 months and formally ends with the removal of the orthodontic braces; patients are then instructed on the use of a retainer to maintain long-term stability. Photographs, radiographic studies, and dental models are obtained on debanding of the orthodontic braces and at 1 year after completion of treatment. Ideally, when possible, patients should have yearly follow-up examinations for an extended period to assess long-term results.

Prosthodontic and Restorative Phase

The final phase includes placement of dental implants, prosthetic and periodontal treatment, and any dental restorations to improve the final dental aesthetics. After the dentoskeletal correction is completed, plastic surgery soft tissue procedures can be considered to improve the final overall facial aesthetics.⁷²

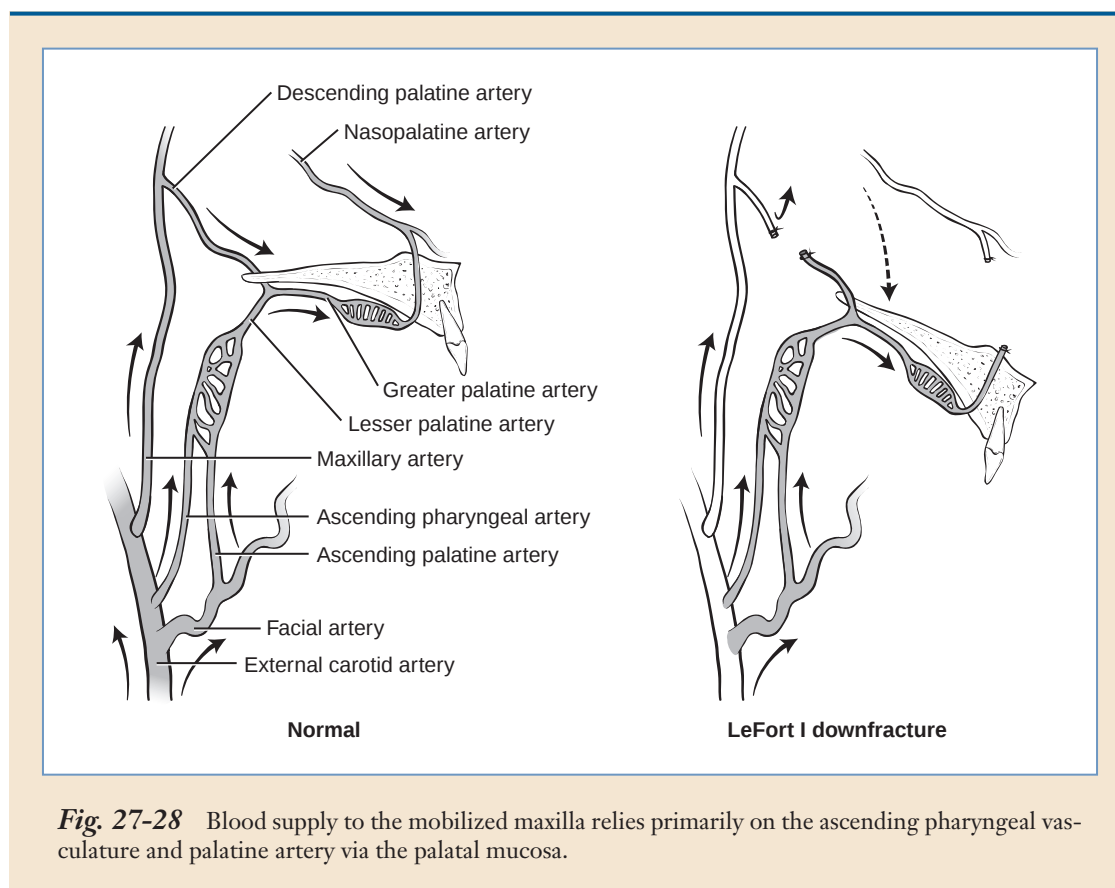
SURGICAL DETAILS

The elements of the facial skeleton can be repositioned, redefining the face through various well-established osteotomies: (1) LeFort I osteotomy, (2) LeFort II osteotomy, (3) LeFort III osteotomy, (4) maxillary segmental osteotomies, (5) sagittal split osteotomy of the mandibular ramus, (6) vertical ramal osteotomy, (7) inverted L and C osteotomies, (8) mandibular body segmental osteotomies, and (9) mandibular symphysis osteotomies.^{1,73-78} The vast majority of maxillofacial deformities can be managed with three basic osteotomies: (1) the midface with the LeFort type I osteotomy, (2) the lower face with sagittal split ramal osteotomy of the mandible, and (3) horizontal osteotomy of the symphysis of the chin.

Midface

Various osteotomies are used to correct midfacial deformities; the choice of a particular procedure depends on the specific deformity. The LeFort osteotomies of today are named after the three classic lines of weakness of the facial skeleton described by René LeFort in 1901. Complete craniofacial disjunction by the LeFort III osteotomy allows the surgeon to alter the orbital position and volume, the zygomatic projection, and the position of the nasal root; it also alters the frontonasal angle and lengthens the nose and position of the maxilla. The LeFort II osteotomy allows the surgeon to alter the nasomaxillary projection without altering the orbital volume and zygomatic projection. The LeFort I osteotomy also allows correction primarily at the occlusal level affecting the upper lip position, the nasal tip and alar base region, and the columellar-labial angle without altering the orbitozygomatic region. Nevertheless, it is important to remember that these standard LeFort osteotomies must be frequently modified to address the specific clinical situation. For example, the standard LeFort I osteotomy must be modified to include a portion of the body of the zygoma when the lower maxillary deficiency is accompanied by inadequate zygomatic projection, but the orbit itself does not need to be altered as it would be in a LeFort III osteotomy. LeFort II and III osteotomies are generally part of the treatment plan in the major craniofacial dysostosis syndromes and are discussed elsewhere in this book. For most midfacial maxillofacial deformities encountered, the LeFort I osteotomy and its variations are adequate.

Deformities of the maxilla can occur in all three planes, and in many patients will frequently occur simultaneously in multiple planes. Even when the primary deformity is in the mandible, including a maxillary repositioning osteotomy, in addition to mandibular surgery, will in many cases improve the postoperative skeletal stability and aesthetic outcome. With its technical ease and dependability of outcome, the LeFort I osteotomy has become the principal procedure used to resolve virtually the full range of anteroposterior, vertical, transverse, and rotational deformities that surgeons may see. Once sectioned, the maxillary dentoalveolar segment can be repositioned to correct occlusal coordination virtually in any direction as a single unit, and when needed, it can be further subsectioned to differentially move the individual elements in all three planes. However, the multiple-segment LeFort I osteotomy increases the complexity and risks of the surgical procedure.



With maxillary osteotomies, an understanding of the vascular blood supply to the mobilized maxilla is crucial (Fig. 27-28). The arterial blood supply to the maxilla is derived from the following four primary sources: (1) the descending palatine branch of the maxillary artery, (2) the ascending palatine branch of the facial artery, (3) the anterior branch of the ascending pharyngeal artery from the external carotid, and (4) the alveolar branches of the maxillary artery. With complete mobilization of the maxilla, the descending palatine vessels are frequently disrupted, and the mobilized maxilla derives its vascularity from the remaining sources, primarily the ascending palatine and pharyngeal vessels.⁷⁹⁻⁸¹

LeFort I Surgical Details

An external reference mark is made by tattooing the medial canthus with methylene blue dye, and the vertical distance is measured to the maxillary archwire by the lateral incisor. (Alternatively, some surgeons prefer to use a K-wire placed at the nasion or an internal bony reference mark with a drill hole before the osteotomy.) This measurement is used to vertically position the maxilla (Fig. 27-29).

Exposure to the midfacial facial skeleton is accomplished through an intraoral labial-buccal incision above the attached gingiva from first molar to first molar, leaving an adequate mucosal edge (a minimum of 5 to 8 cm) to facilitate final closure (Fig. 27-30).

The subperiosteal dissection is then directed superiorly exposing the anterior maxillary wall, identifying the inferior orbital foramen, exposing the lateral zygomatic-maxillary buttress, the body of the zygoma, and the anterior portion of the zygomatic arch.

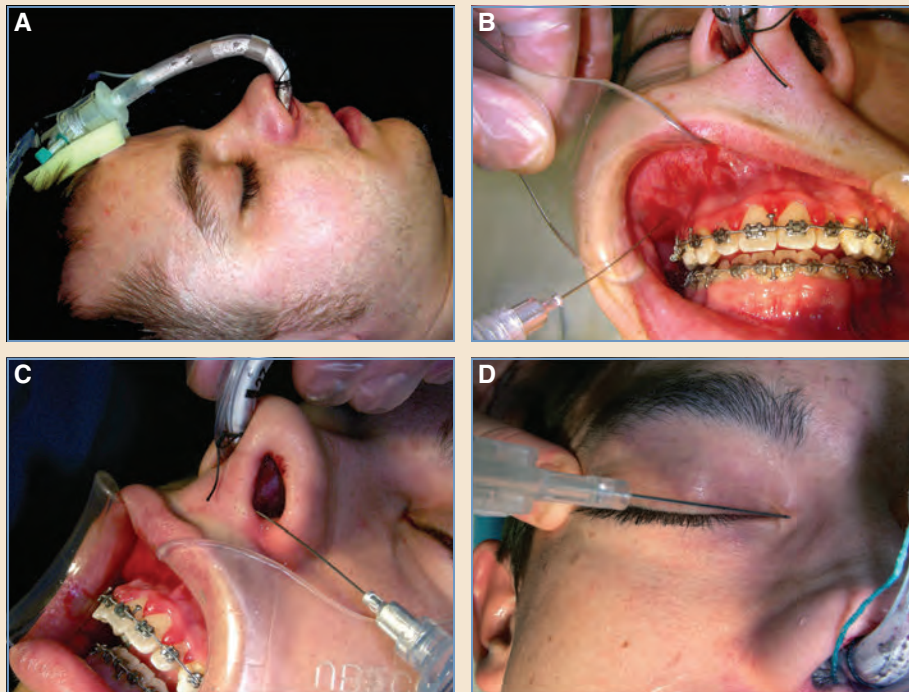


Fig. 27-29 LeFort I procedure. **A**, Nasotracheal intubation is secured at the septum and scalp without distorting the ala. **B** and **C**, Infiltration with a local vasoconstricting agent. **D**, Methylene blue is used to mark the medial canthus for vertical reference.

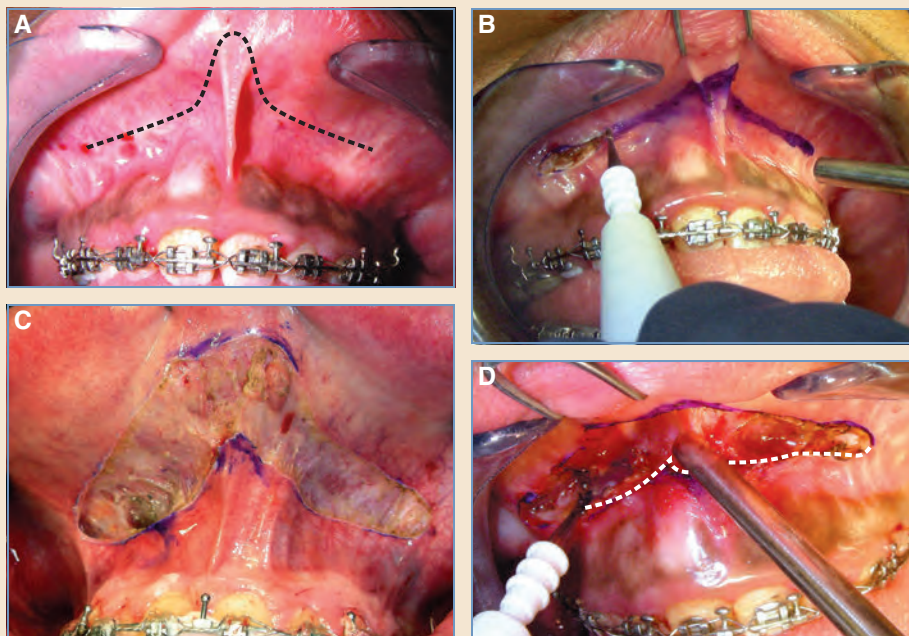


Fig. 27-30 **A** and **B**, The incision should be made well above the attached gingiva, sparing the frenulum, from premolar to premolar. **C** and **D**, The incision should then be made deeper along the inferior edge to avoid entering the nasal mucosa centrally.

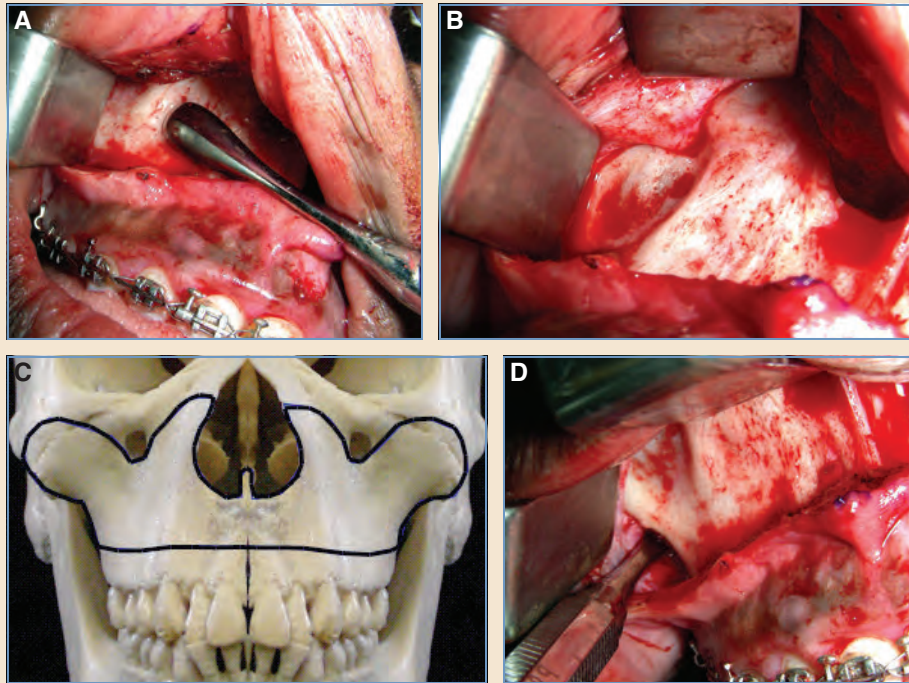


Fig. 27-31 A, Subperiosteal dissection. B, Exposure of the supraorbital nerve. C, Area of skeletal exposure. D, Exposure of the pterygopalatine region.

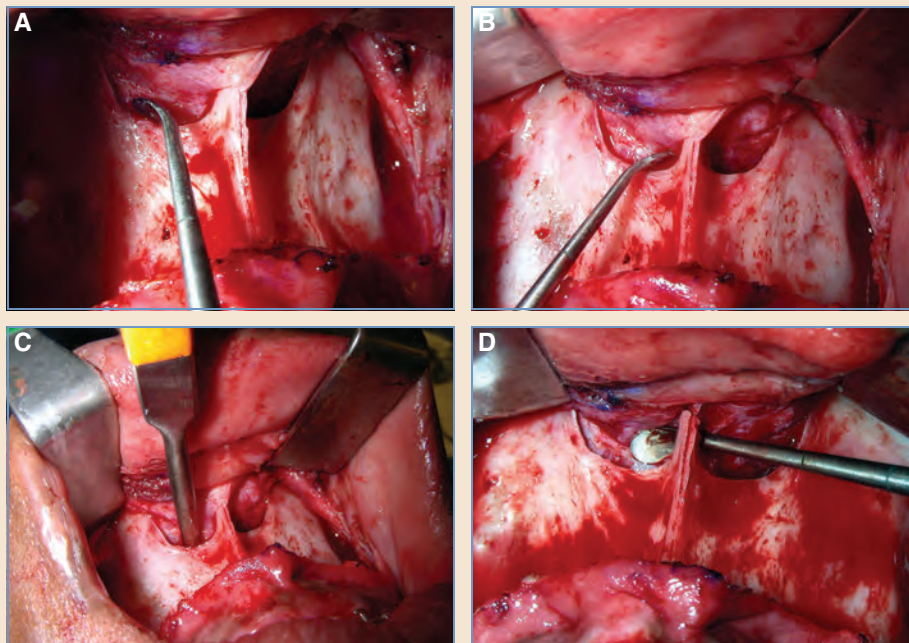


Fig. 27-32 A and B, Exposure of the piriform aperture. C, Exposure of the lateral nasal wall inferior to the turbinate and the nasal floor to the junction of the hard and soft palate. D, Exposure and separation of the anterior nasal spine from the septum.

The superior exposure must be sufficient to accommodate the fixation plates. The dissection continues posteriorly to the pterygopalatine region. Dissection is then directed medially to expose the nasomaxillary buttress, piriform aperture, and anterior nasal spine. The intranasal dissection is then carried out exposing the nasal floor, the lateral nasal wall below the inferior turbinate, and medially the septum and vomer. The dissection should continue posteriorly to the junction of the hard and soft palate (Figs. 27-31 and 27-32).

With the skeletal exposure complete, the planned osteotomy is marked noting the tooth root apices and infraorbital foramen. There are many variations on the standard LeFort osteotomy, and as previously stated, each osteotomy must be tailored to the individual patient (Figs. 27-33 and 27-34). A reciprocating saw is used to complete the horizontal maxillary osteotomies, proceeding from the lateral nasal wall across the anterior maxillary wall and through the posterior lateral maxillary wall. If needed, the reciprocating saw can be redirected from lateral to medial to facilitate completion of the osteotomy.

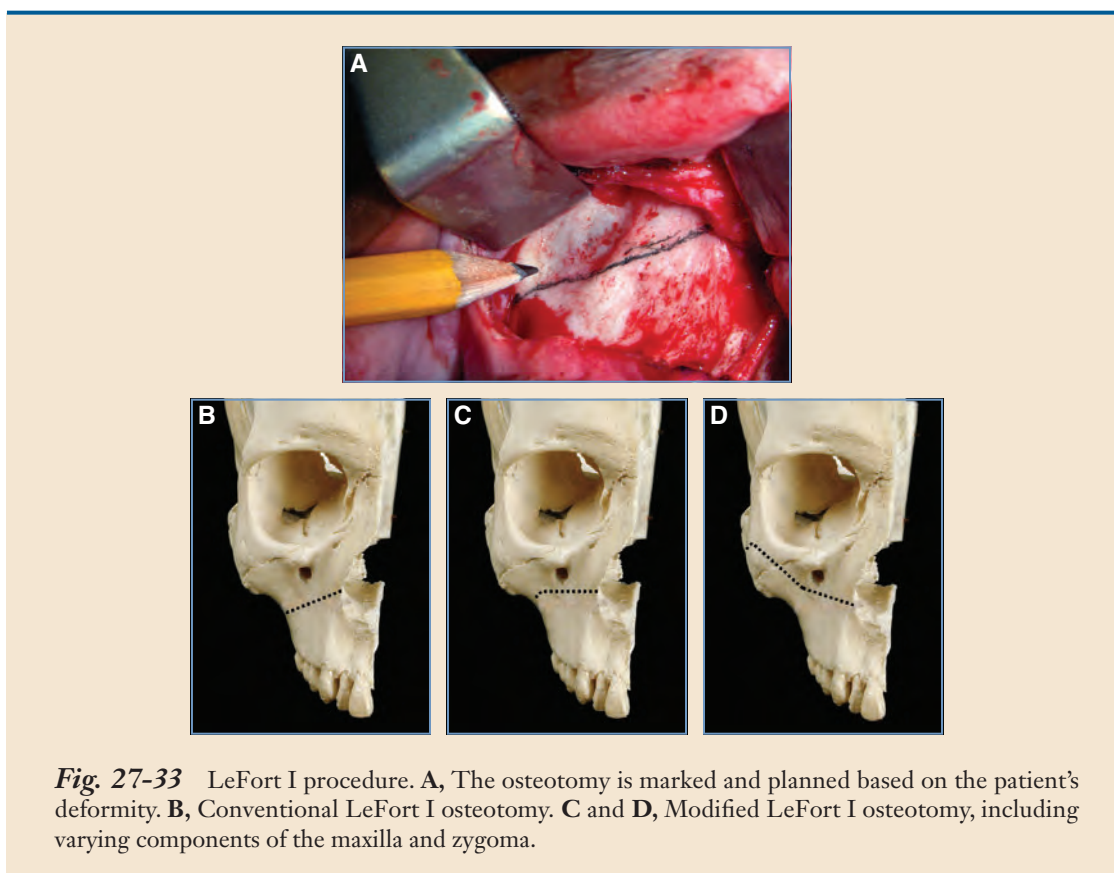


Fig. 27-33 LeFort I procedure. **A**, The osteotomy is marked and planned based on the patient's deformity. **B**, Conventional LeFort I osteotomy. **C** and **D**, Modified LeFort I osteotomy, including varying components of the maxilla and zygoma.

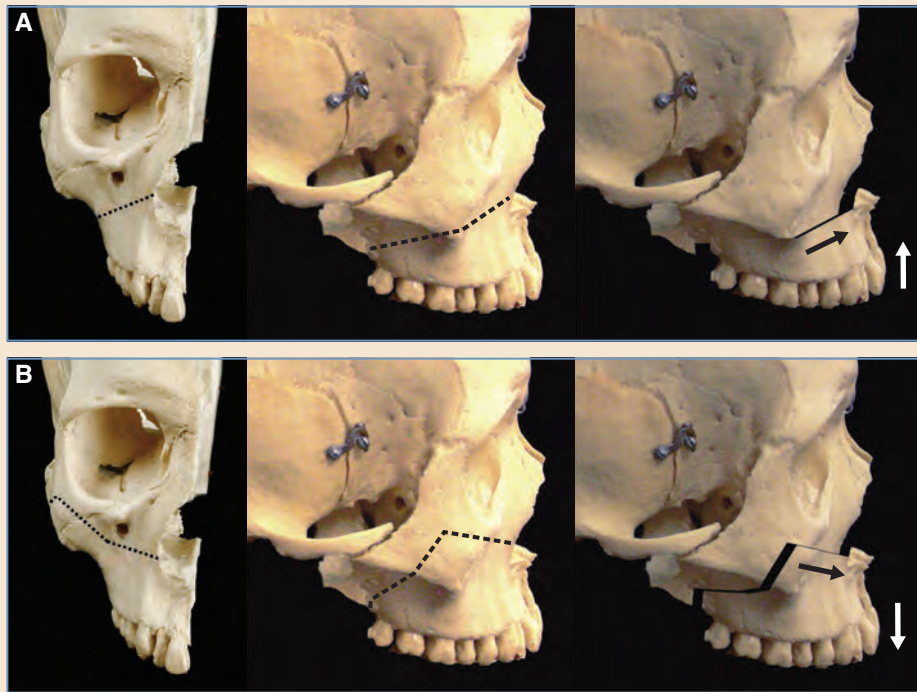


Fig. 27-34 A, Conventional LeFort I osteotomy. B, Modified LeFort I osteotomy. The difference between these two procedures is shown. By varying the osteotomy design and angulation, the anterior vertical height and dental display can be controlled. With a conventional LeFort I osteotomy, increasing the anterior vertical height requires increasing the osteotomy gap and the need for bone grafting to ensure stability.

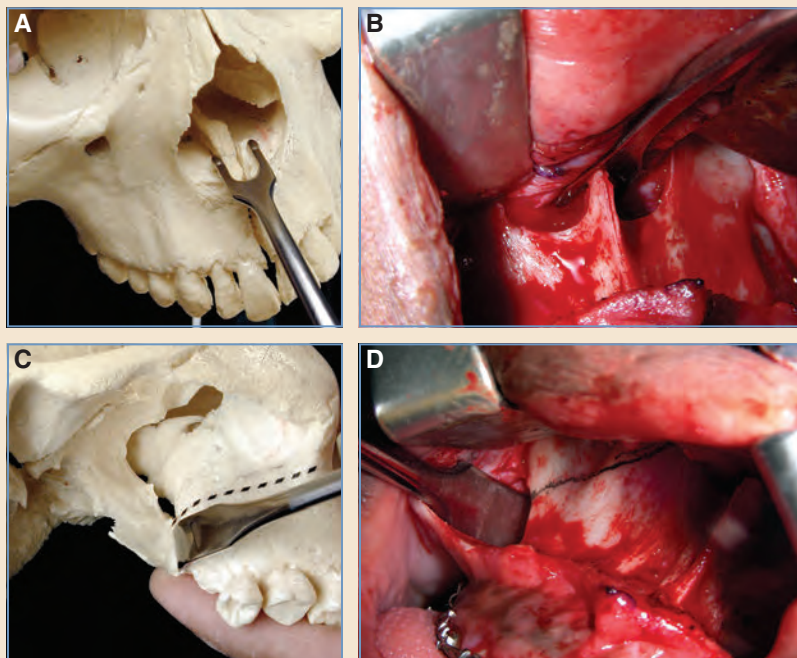
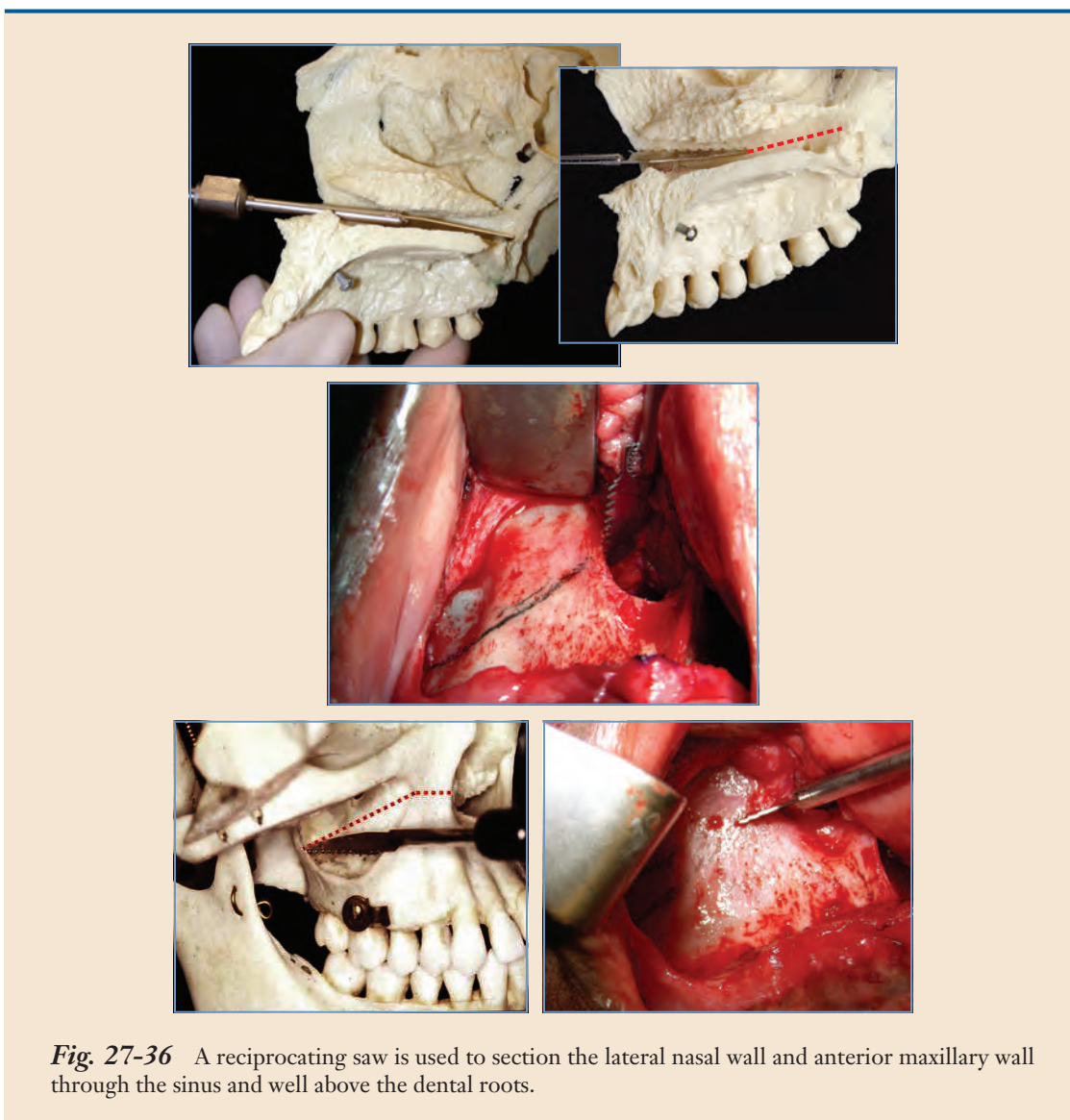


Fig. 27-35 A and B, The septum is separated from the vomer from the anterior nasal spine to the posterior nasal spine. C and D, The pterygoid plate is separated from the maxillary-palatine bone.

The nasal septum and vomer are then separated from the maxillary crest with a septal osteotome that is directed downward and posteriorly. Alternately, the septal osteotomy can be accomplished with a reciprocating saw that is directed away from the side with the nasal intubation to avoid transecting the nasotracheal tube. Finally, a curved osteotome is used to separate the pterygoid plate from the maxillary tuberosity. The pterygopalatine disjunction osteotomy should be positioned parallel to the occlusal plane and below the maxillary osteotomy to avoid injuring the internal maxillary artery. A finger placed behind the maxillary tuberosity can easily verify the separation. Some surgeons prefer to use an oscillating saw instead of the osteotome (see Figs. 27-33 through 27-36).



After the osteotomy is completed, the maxilla is downfractured (Fig. 27-37). This can typically be achieved with digital pressure alone by placing the thumbs at the base of the piriform aperture. If there is any significant resistance, it is preferable to revisit the osteotomy sites with the reciprocating saw and an osteotome and then immediately proceed with Rowe disimpaction forceps. The use of significant force to accomplish the downfracturing can lead to unfavorable fractures. The areas that are typically incomplete are the posterior aspect of the lateral nasal walls and the pterygopalatine disjunction. As the maxilla is mobilized inferiorly, any adherent nasal floor mucosa should be simultaneously elevated. Once the downfracture is completed, the maxilla is mobilized from side to side and then anteriorly in the sagittal direction with the aid of Tessier retromaxillary levers or Rowe forceps and releasing any adherent soft tissue from the posterior aspect behind the tuberosity. The maxilla should be so completely mobilized that it can be passively placed in its final position with a pair of forceps (Figs. 27-38 and 27-39).

With the maxilla downfractured and mobilized, a prefabricated occlusal splint is then ligated to the maxillary archwire, and the maxilla and mandible are placed in maxillary-mandibular fixation either with wire or dental elastics. The maxillary-mandibular complex is allowed to rotate within its arc of rotation, with the mandibular condyle seated in its glenoid fossa. The maxilla is then brought into the desired anterior vertical position based on the amount of dental display desired from the preoperative analysis. This is confirmed by the vertical measurement from the previously marked reference point. Any areas of bony interference should be removed, along with the nasal septum, to prevent septal buckling. The removal of bone should be done selectively to

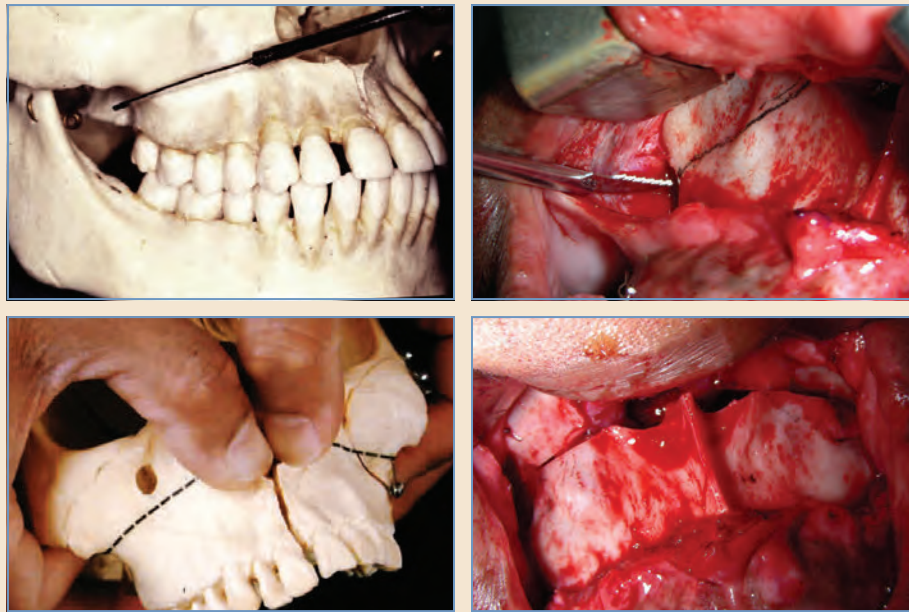


Fig. 27-37 A reciprocating saw is used to section the lateral nasal wall and anterior maxillary wall through the sinus and well above the dental roots.

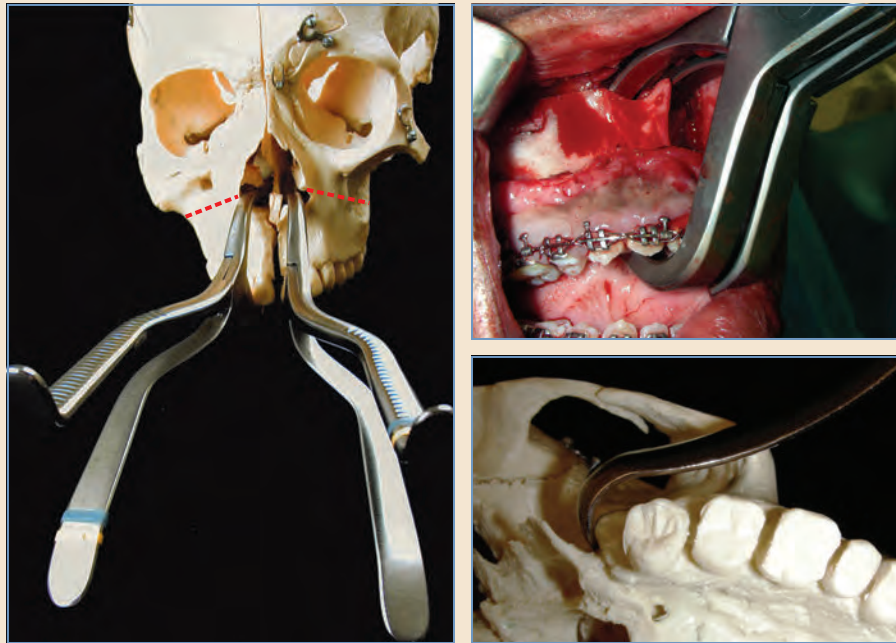


Fig. 27-38 After the downfracture is completed, Rowe and retromaxillary forceps can be used to further mobilize the maxilla.

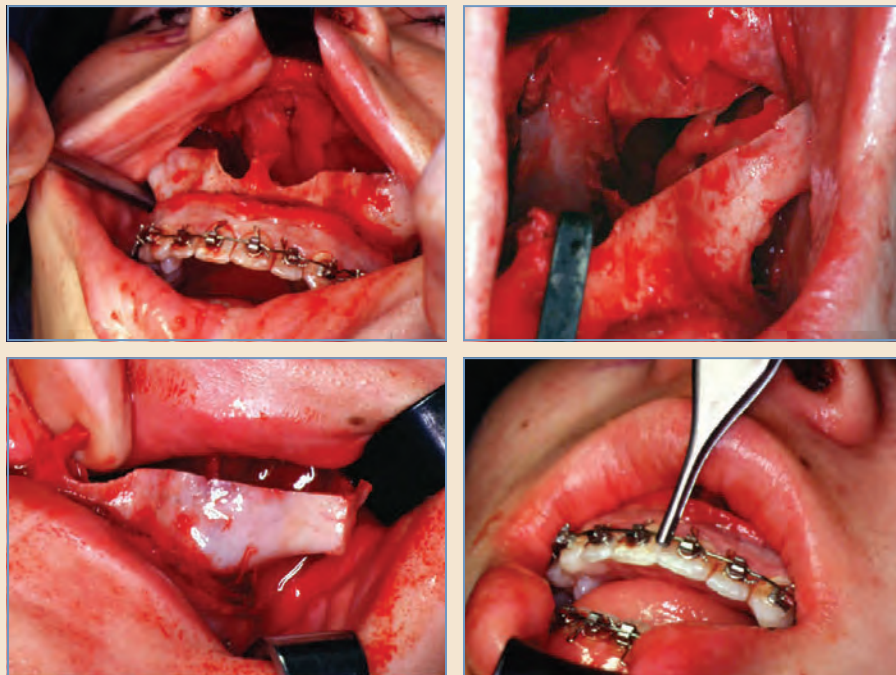


Fig. 27-39 The maxilla should be mobilized so that forceps can easily reposition the maxilla alone without tension. Frequently the soft tissue may need to be further freed.

allow maximal bony contact for stability. When the maxilla is in the desired position, it is stabilized with plates and screws at the medial and lateral buttresses. The maxillary-mandibular fixation is then released, and the occlusion is verified so that the mandible passively closes directly into the splint without repositioning. If there is any question that the condyle was not properly seated at the time of fixation, the osteosynthesis must be redone. If there is significant inferior maxillary repositioning, either autologous bone or alloplastic hydroxyapatite should be used for interpositional buttress grafts (Figs. 27-40 through 27-43).

After the maxilla is rigidly fixed and the occlusion is satisfactory, the operative wounds are closed. The septum should be fixed in the midline and an alar cinch suture placed for a wide-flaring ala if needed. At the time the vestibular incision is closed, cheiloplasty with V-Y advancement or any number of variations can be considered.

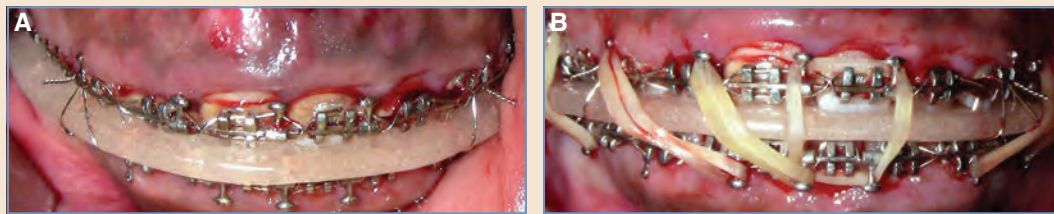


Fig. 27-40 A, A surgical splint fabricated from the model surgery is wired to the maxillary dental archwire. B, The maxilla and mandible are then placed in fixation with dental elastics.

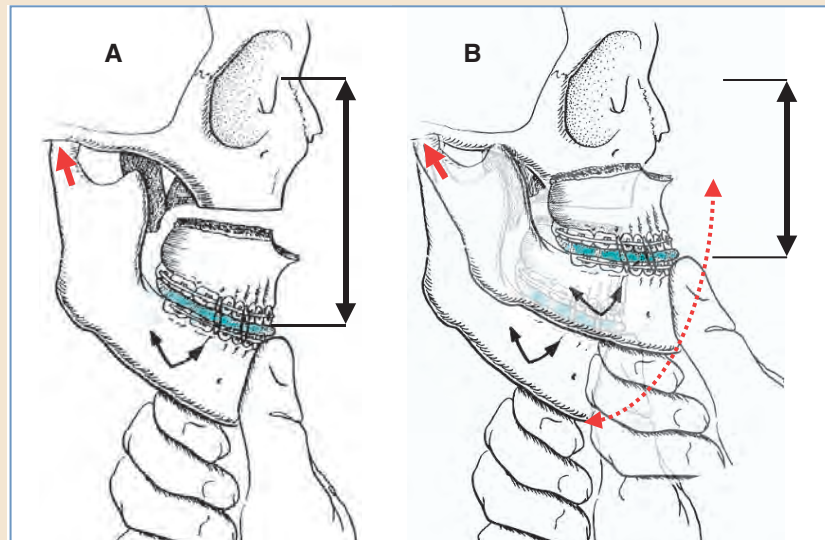


Fig. 27-41 The vertical height is measured from the medial canthus marked with methylene blue to the incisor archwire before the maxilla is downfractured. With the condyles seated within the glenoid fossa, the vertical position of the maxilla is determined based on the desired anterior facial height and dental display. A, Increase in vertical length and incisor show. B, Decrease in anterior vertical length with less incisor show (impaction).

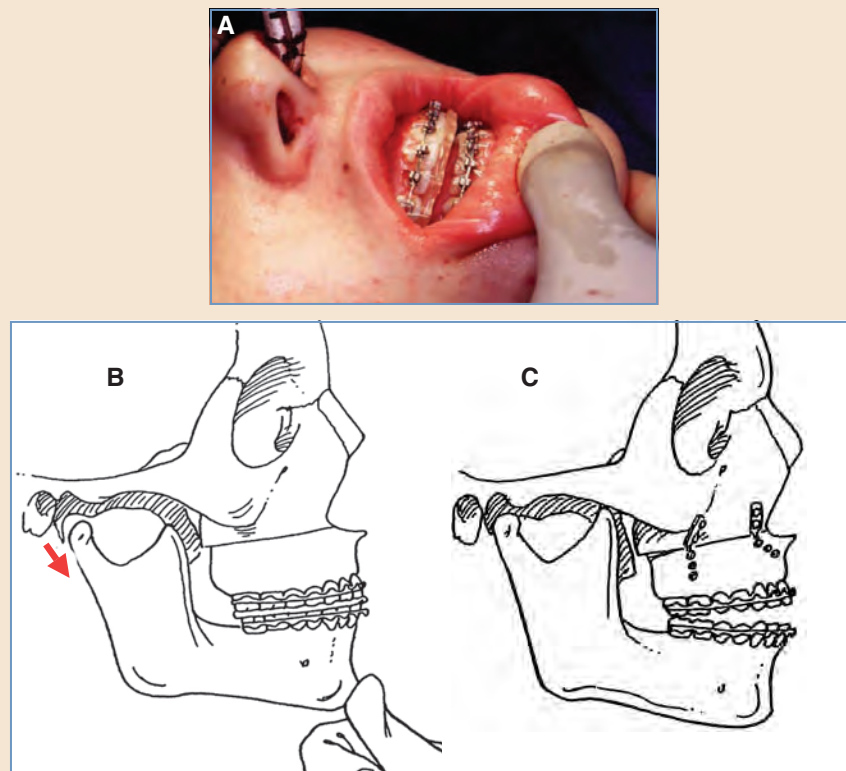
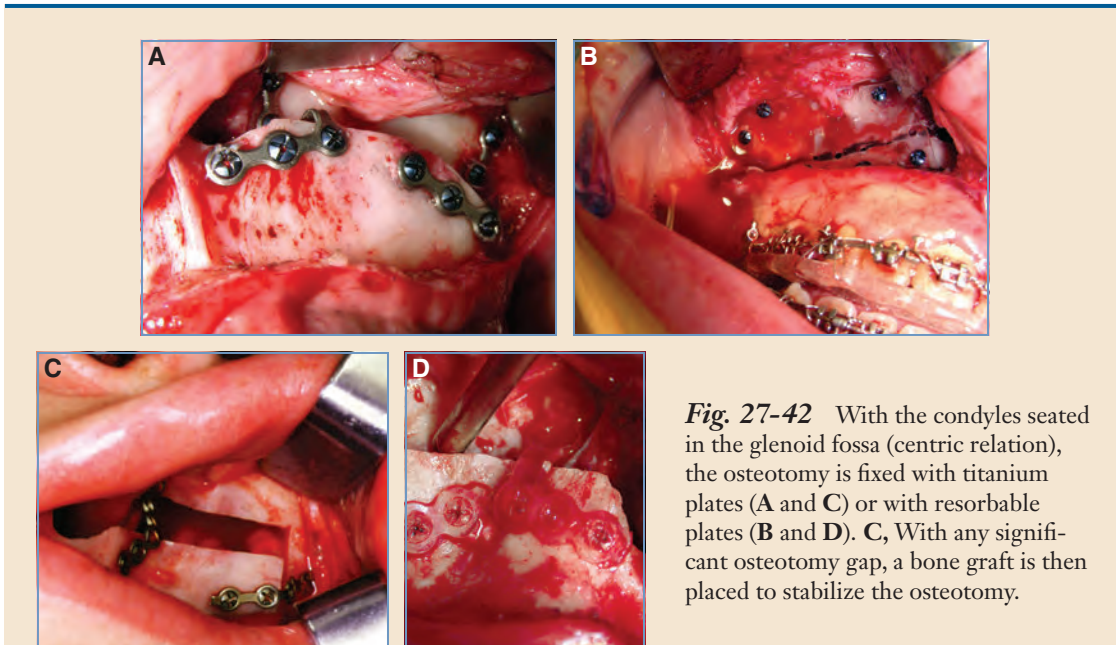


Fig. 27-43 A, The maxillary-mandibular interdental fixation is removed, and the occlusion is checked to confirm whether the mandible directly comes into the splint with the condyles seated. B and C, If the condyles are not properly seated at the time of fixation, the malocclusion will become readily evident (anterior open bite). The plates will need to be removed, and the maxilla and mandible are placed in elastic fixation. The maxilla is replated with the condyles seated within the glenoid fossa.

Lower Face

For the lower face, various osteotomies are used to correct mandibular deformities; which one to use depends on the particular deformity. Today the sagittal split ramal osteotomy is the workhorse for correcting the vast majority of cases of mandibular retrognathism and prognathism. In extreme cases of mandibular prognathism, some surgeons prefer the intraoral vertical osteotomy (IVO) or inverted-L osteotomy. In cases of mandibular advancement when the mandibular rami are hypoplastic and cannot be sagittally split, the inverted-L and C osteotomy with bone grafts are preferred. Deformities of the chin can exist independent of mandibular deformities and can be abnormally proportioned in and of themselves without occlusal involvement. Although alloplastic chin implants have been most commonly used for correction of minimal sagittal chin deficiencies, the horizontal osteotomy of the symphysis (osseous genioplasty) is a far more versatile procedure. The chin can be repositioned in multiple planes, allowing correction of significant sagittal and vertical deformities whether of deficiency (microgenia) or excess (macrogenia), along with asymmetrical conditions.

With mandibular osteotomies, the surgeon must be cognizant of the course of the inferior alveolar nerve from its entrance at the mandibular foramen on the medial aspect of the ramus to its emergence from the mental foramen between the first and second premolars to minimize neurosensory deficits. Regarding the vertical position, the mandibular foramen typically lies approximately 8 cm inferior to the lingula, and the lingula itself is approximately 5 cm above the occlusal plane. Using the sigmoid notch as a reference point, the foramen is approximately 20 cm inferior. Regarding the anterior to posterior relationship, the foramen is located 20 mm from the anterior mandibular ramal border, a depth that is approximately two thirds of the total mandibular ramal width. The canal then courses within the mandible, measuring 2 to 2.5 mm in diameter. Its lowest point from the inferior mandibular border is in the region of the first and second molars, approximately 7.5 cm, before it continues anterior and superior to its emergence from the mental foramen, where it is approximately 8 mm from the inferior border. At the mental foramen, the canal extends caudally before emerging. Regarding the transverse position of the canal within the mandible, the canal is most superficial in the region of the third molar, approximately 2 mm from the buccal plate. In the region of the first molar, it is 4 mm from the buccal plate.^{82,83}

Sagittal Split Osteotomy of the Mandibular Ramus

The ascending ramus is palpated, and the mucovestibular incision is placed well lateral to the posterior dentition to allow an adequate medial mucosal edge for final closure. The incision typically begins approximately 1 to 2 cm above the occlusal plane and continues to the region of the first molar (Fig. 27-44).

The incision then continues through the periosteum, exposing the lateral border of the posterior body, angle, and ascending ramus. The inferior mandibular border is exposed with an inferior border stripper so that a channel retractor can be easily placed. The anterior ramal border is partially stripped of the temporalis muscle, and a retractor is placed superiorly onto the coronoid process. The medial portion of the ramus is elevated with a curved periosteal elevator beginning well above the occlusal plane proceeding from superior to inferior so that the inferior alveolar neurovascular bundle is safely reflected medially. In elevating the medial ramus, care must be taken to remain subperiosteal and to recall the anatomic topography with the lingula and the relationship of the mandibular foramen (Fig. 27-45).

With the exposure complete, a medial ramus retractor or a curved elevator is then placed to protect the neurovascular bundle. The horizontal osteotomy of the medial ramus is made first with either a Lindemann side-cutting burr or a reciprocating saw starting well above and posterior to the mandibular foramen. The osteotomy is deepened to approximately half the thickness

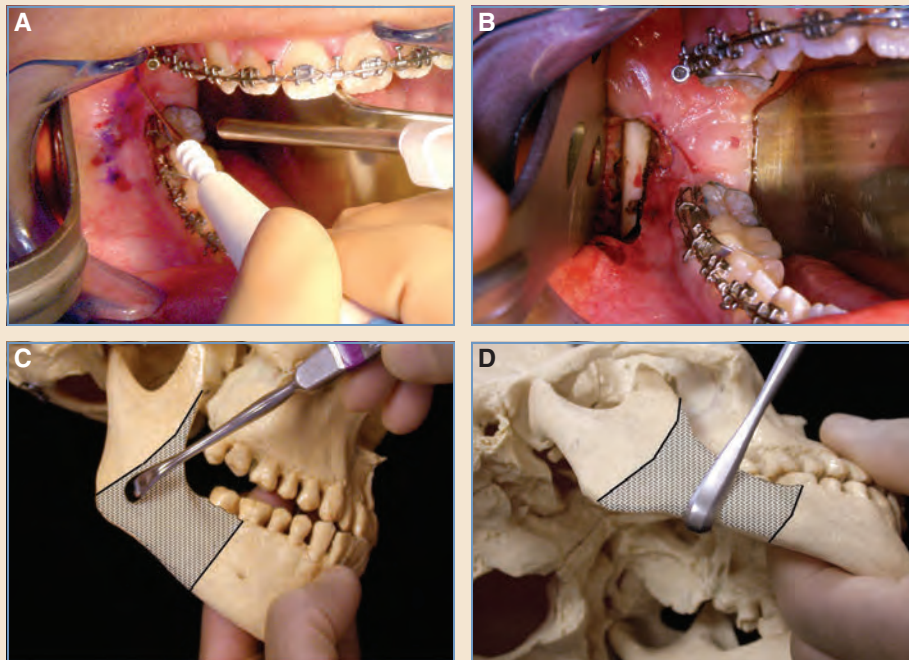


Fig. 27-44 BSSO of the mandible. **A**, The incision is made well lateral to the attached gingiva in the molar region. **B**, The mandibular ramus is exposed in the subperiosteal plane. **C**, The lateral ramus, angle, and body are exposed. **D**, The inferior border is exposed.

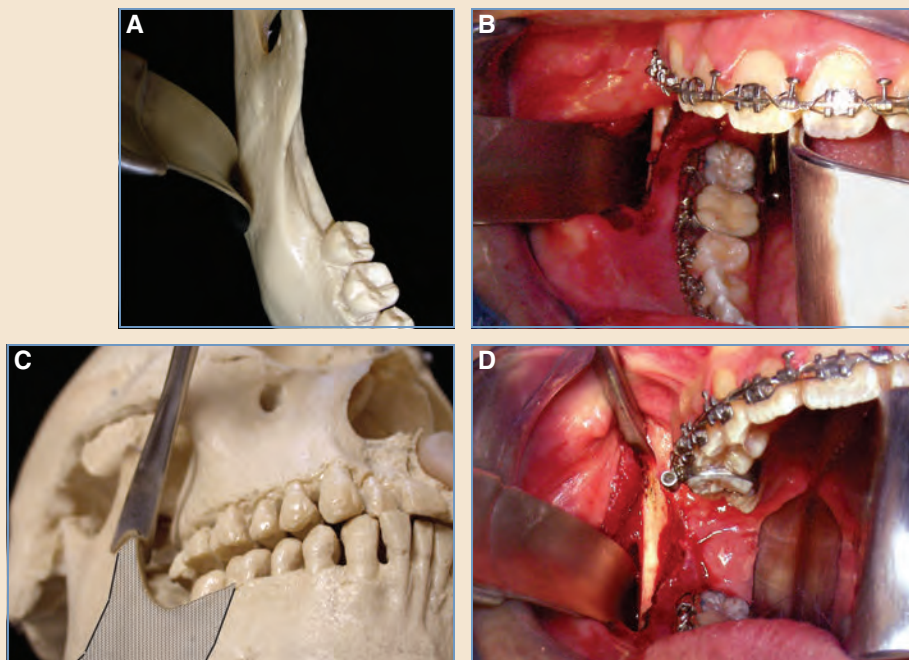


Fig. 27-45 **A** and **B**, A channel retractor is placed to support the mandible. **C** and **D**, The anterior ramal border and coronoid process are exposed to the insertion of the temporalis muscle.

of the ramus and obliquely made so that the osteotomy can be continued inferiorly. A reciprocating saw is used to continue the osteotomy laterally along the external oblique ridge to the region between the first and second molars. The tip of the saw should only penetrate the anterior cortex, and the osteotomy should be placed as far lateral as possible to protect the nerve. The saw is then repositioned to make the vertical osteotomy through the buccal cortex, typically between the second and first molars. Care must be taken to ensure that the vertical osteotomy continues through the inferior mandibular border to the medial lingual cortex. The remainder of the osteotomy is completed with a series of osteotomes, beginning from mesial to distal, approaching the horizontal osteotomy of the mandible. The osteotomes are directed laterally toward the buccal cortex to avoid transecting the inferior alveolar nerve. A three-point spreader can be placed to begin the separation, which can be completed with an osteotome at points of resistance. The course of the inferior alveolar nerve should be entirely within the distal tooth-bearing segment. If it is not, the nerve must be mobilized from the proximal segment. The separation must be complete so that the mesial tooth-bearing segment will move independent of the distal condylar segment and can be repositioned into final occlusion with little resistance. The osteotomy on the contralateral side is then carried out (Figs. 27-46 through 27-49).

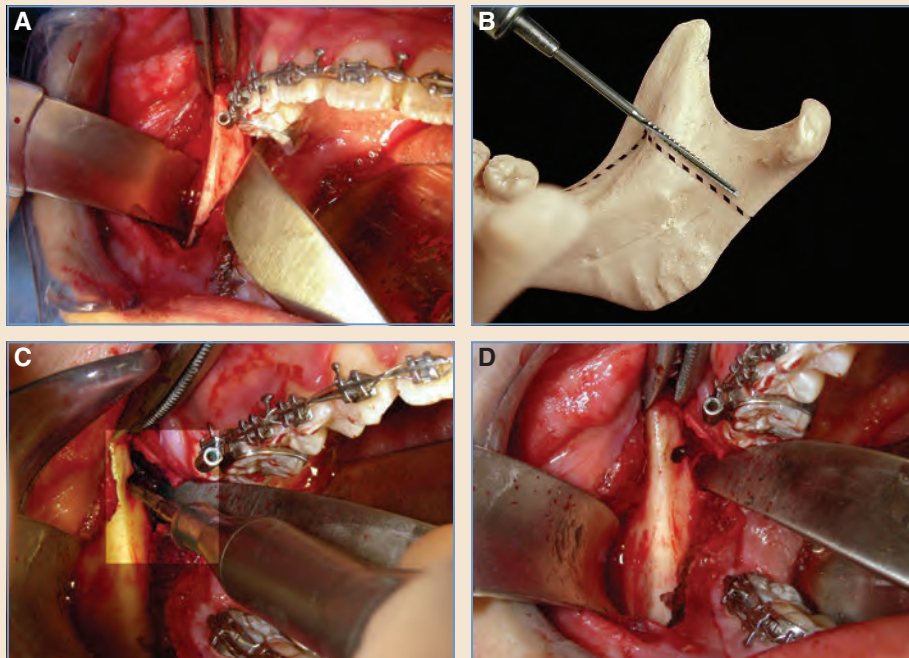


Fig. 27-46 A, A medial ramus retractor is placed to protect the nerve. B-D, A horizontal osteotomy is made with a Lindemann burr, or a reciprocating saw can be used.

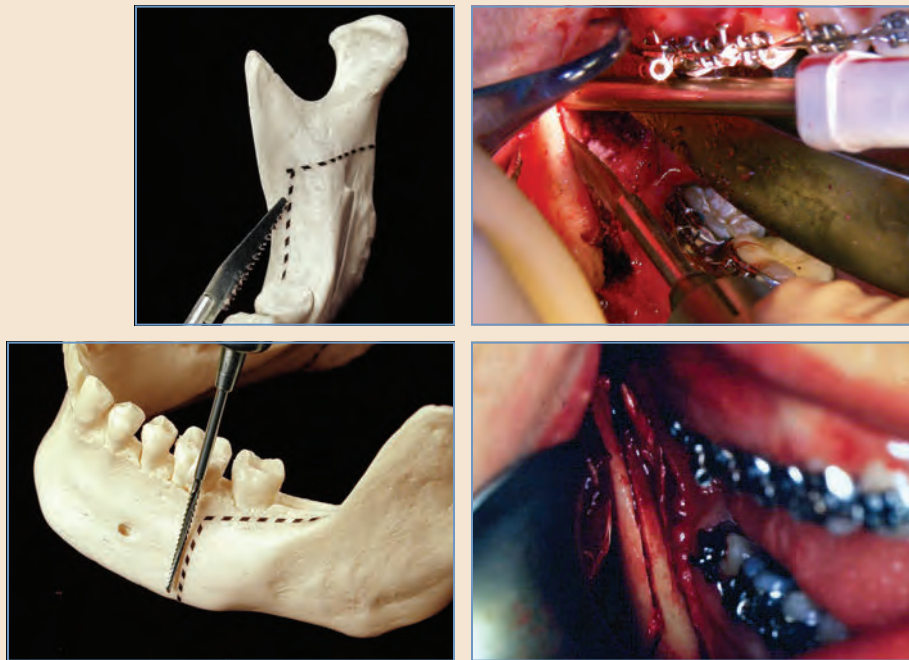


Fig. 27-47 A reciprocating saw is then used along the external oblique ridge and continued to the region of the first and second molars, where a vertical osteotomy is made. The vertical osteotomy can be made with either a Lindemann burr or a reciprocating saw.

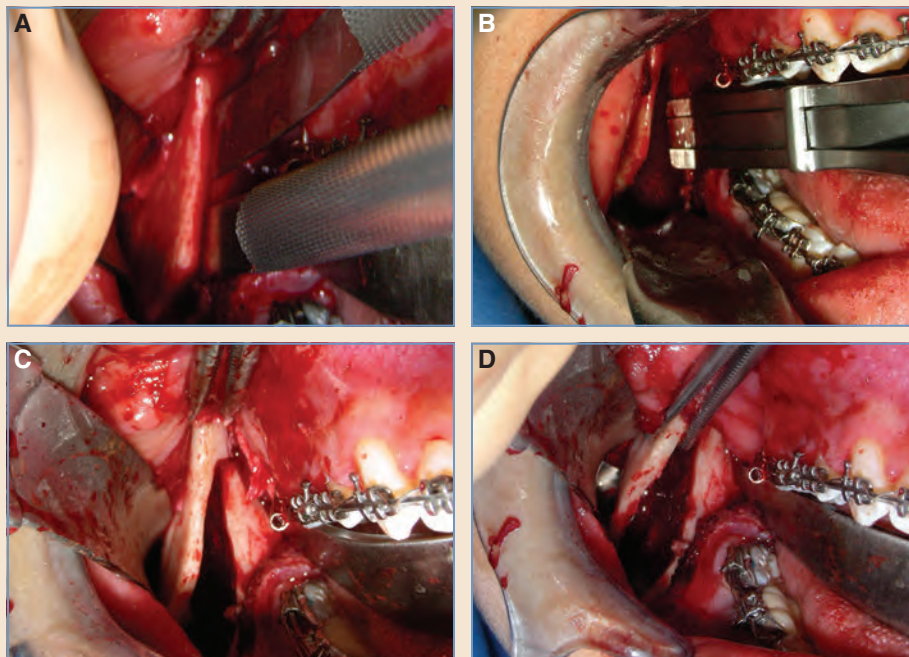


Fig. 27-48 BSSO of the mandible. **A**, With a series of osteotomes, the corticotomy is converted to an osteotomy. The osteotomes should be directed laterally along the medial surface of the lateral or buccal cortex to minimize injury to the nerve. **B**, A ramal spreader is then introduced, and the medial and lateral cortices are separated, complete with the nerve within the medial component. **C** and **D**, The lateral ramal component should be completely freed and independent of the distal dental arch.

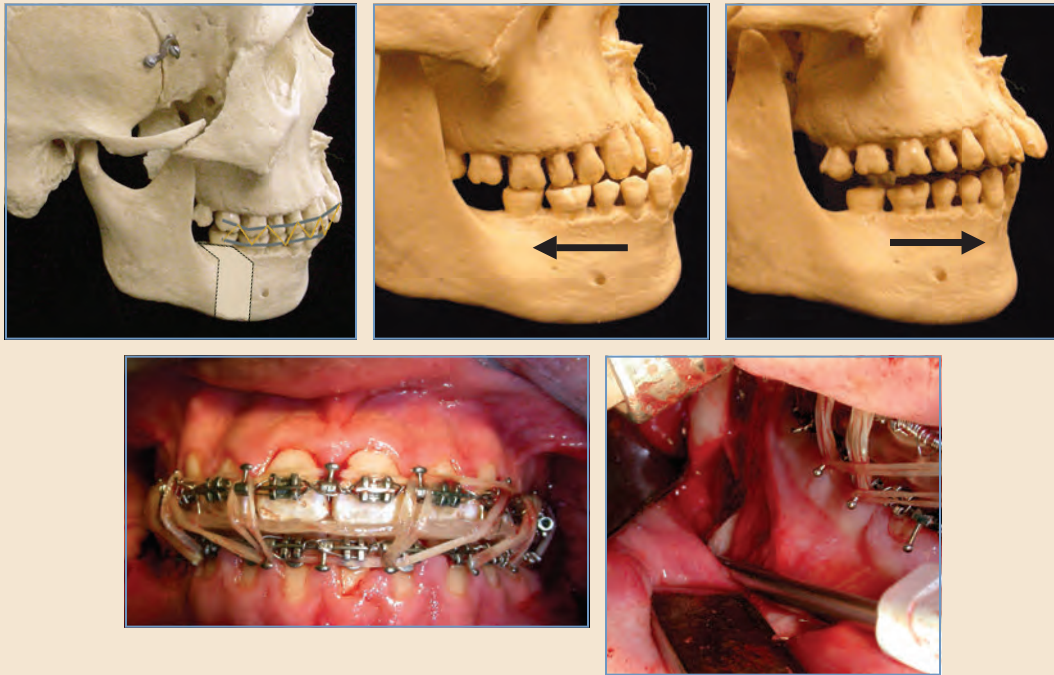


Fig. 27-49 BSSO of the mandible. The mandible is then either advanced or set back by placing it within the prefabricated surgical splint and maxillary-mandibular dental fixation.

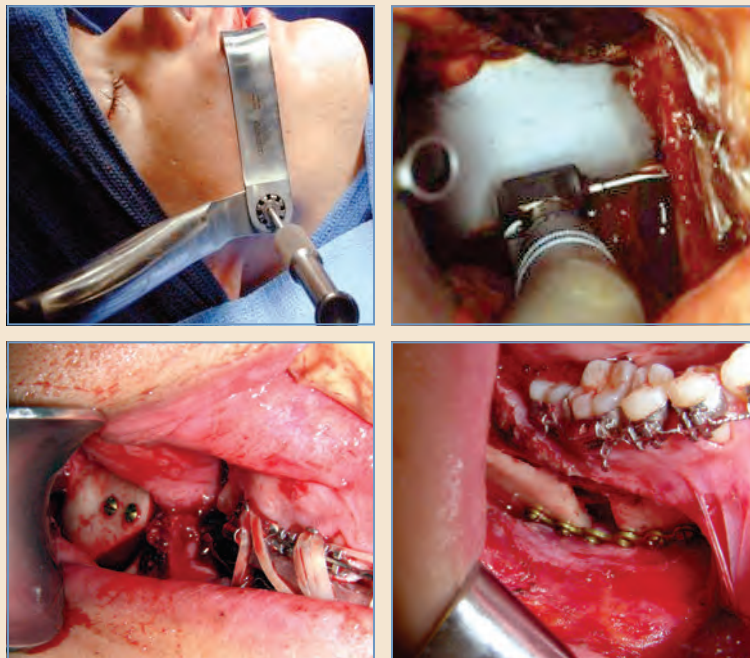


Fig. 27-50 BSSO of the mandible. The repositioned elements are then fixed either with positional screws placed through a transbuccal approach or with titanium plates through an intraoral approach.

The mesial (distal) tooth-bearing segment is placed in maxillary-mandibular fixation with a prefabricated occlusal splint ligated to the maxillary archwire. The condylar distal (proximal) segment is seated within the glenoid fossa. Fixation is typically obtained by placing three bicortical screws in an inverted-L pattern or with an inferior border monocortical plate and a single positional bicortical screw. Fixation can be entirely transoral with a contraangle drill and screw-driver system or a transbuccal trocar approach. It is important to note that the bicortical screws are positional screws used to “loosely” approximate the bony segments and are not lag screws. If the screws are tightened down, the proximal segment may be laterally displaced out of the glenoid fossa, and there is some risk of a “crush” injury to the nerve. The maxillary-mandibular fixation is released and the occlusion is checked. If the occlusion is unacceptable, the fixation must be removed and the segments repositioned. If the occlusion is satisfactory, the wounds are then approximated (Fig. 27-50).

Osseous Genioplasty

A labial mucosa incision, typically from canine to canine, is made well above the attached gingiva to allow closure. The mentalis muscle is partially transected from its insertion, and a subperiosteal dissection is carried out. The dissection over the pogonion must be limited in scope so that the dissection continues below the mental foramen and as far lateral as possible to the inferior mandibular border in the region of the second premolar and first molar (Fig. 27-51).

With the exposure complete, the osteotomy is marked a minimum of 5 to 6 mm below the tooth root apices and similarly below the mental foramen. The surgeon must remember that the canine length is approximately 30 mm and that the mental foramen extends caudally before it emerges.

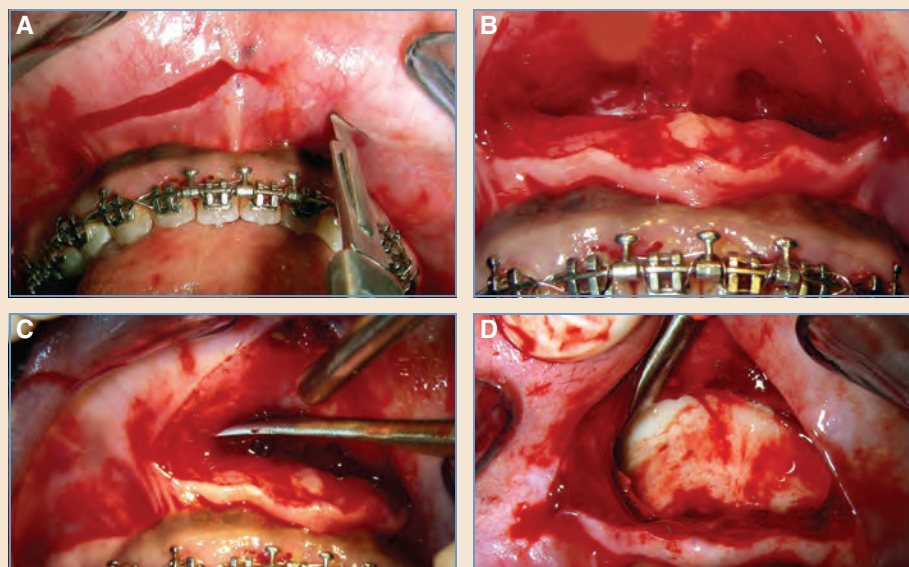


Fig. 27-51 Genioplasty. **A**, The incision is made inferior to the attached gingiva. **B**, The mentalis muscle is then divided, leaving an adequate cuff of muscle for reattachment at the time of closure. **C** and **D**, The symphysis is exposed in the subperiosteal plane.

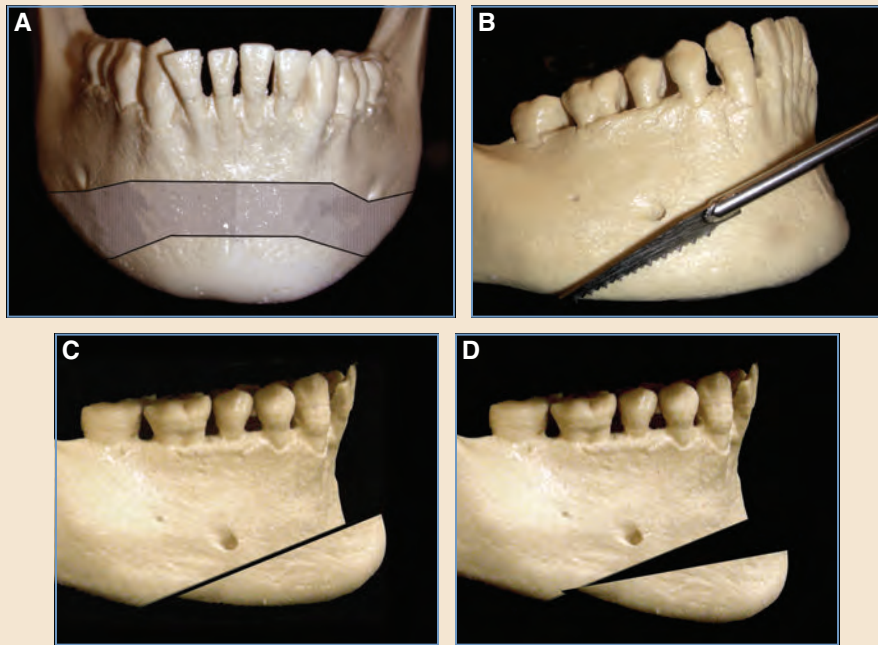


Fig. 27-52 A, The area of exposure is limited to leave a muscular attachment at the symphysis (vascular pedicled genioplasty). B, The osteotomy is made as posterior as technically possible, 5 to 6 mm below the mental foramen. C and D, The chin can be repositioned in all three planes to correct the sagittal, vertical, and transverse components of the chin deformity.

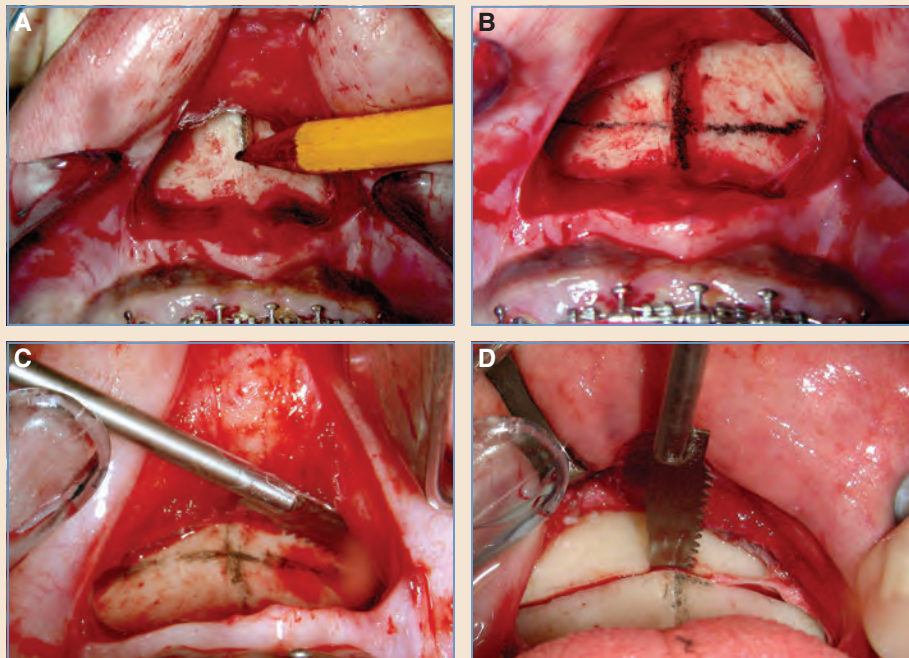


Fig. 27-53 A, The midline is marked. B, The planned osteotomy is then marked well below the dental roots and mental foramen. C and D, The osteotomy is made with a reciprocating saw.

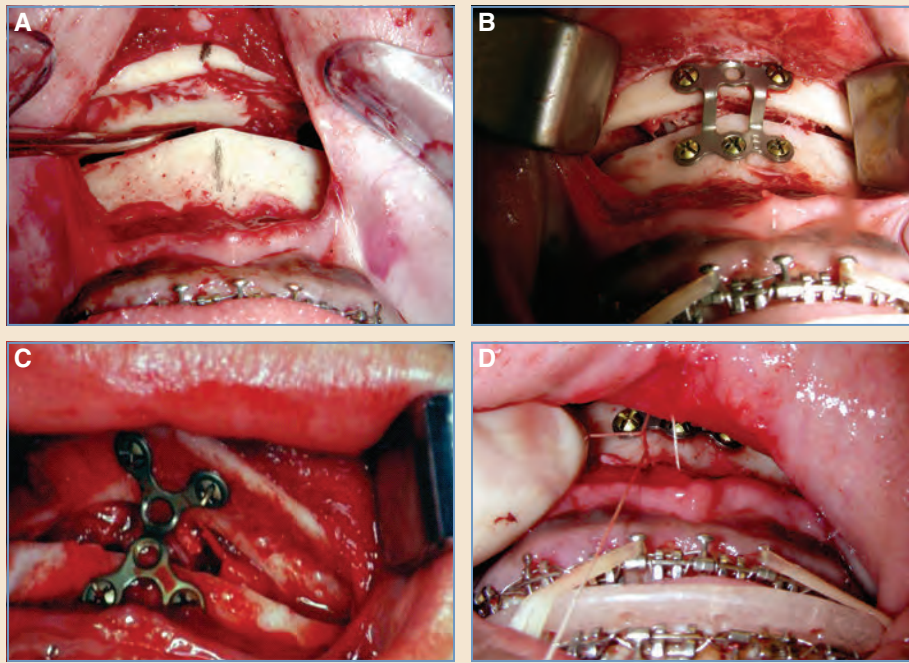


Fig. 27-54 A, The symphysis is mobilized. B and C, Two methods of plate fixation. D, The mentalis muscle is approximated at the time of mucosal closure.

The osteotomy can be varied, depending on the deformity and the planned correction. The midline is scored with an oscillating saw, and the symphyseal osteotomy is made with a reciprocating saw.

If a double osteotomy is planned either for a step advancement or a wedge resection, the distal or inferiorly marked osteotomy should be made first. The lateral inferior border osteotomies must be completed carefully with the reciprocating saw. The inferior segment should easily separate if the osteotomy is complete (Figs. 27-52 and 27-53).

The inferior pedicled osseous segment is then mobilized to the desired position and fixed with plates and screws. A step deformity is typically palpable at the inferior border mandibular osteotomy sites. Although the deformity will become less significant with remodeling, this can be contoured with a guarded rasp (Fig. 27-54). The wound is then closed by approximating the mentalis muscle and overlying mucosa.

Sequencing the Intraoperative Procedure

The sequence of operative procedures is based on the preoperative planning and model surgery. If a single jaw is being repositioned, a final splint is then used to guide the occlusion of the jaw being moved relative to the remaining jaw. If both maxillary and mandibular repositioning osteotomies are planned, an intermediate splint is then used to guide the movement of one jaw relative to the other jaw. Although the maxilla is typically repositioned first, followed by the mandible, the sequence can be reversed with the appropriate intermediate splint at the time of the model surgery. The remaining jaw is then repositioned based on the final splint. After the maxilla and mandible are in their final positions, the chin is assessed, and an osseous genioplasty is performed if necessary.

MANAGEMENT OF DEVELOPMENTAL DENTOFACIAL DEFORMITIES

Mandibular Deficiency

Patients with mandibular deficiency clinically present with a convex facial profile (Figs. 27-55 and 27-56). Although the aesthetic soft tissue analysis of the upper and middle thirds of the face is within an acceptable norm, the lower third of the face is retruded. Along with it, the lower lip is everted with a deep labial mental crease, and because there is lip incompetence, mentalis muscle strain is seen with lip closure. The neck length may be short with an obtuse cervical mental angle, and redundancy of the soft tissue may be present. Dental examination will show an Angle class II malocclusion that is further subdivided into Angle division 1 or Angle division 2 based on the incisor relationship. In Angle class II division 1, the maxillary incisor angulation is within an acceptable range and there is significant overjet. In Angle class II division 2, the maxillary incisors are retroclined, giving the appearance of less overjet, and in addition, there is an associated deep bite and an overaccentuated curve of Spee, resulting in decreased lower facial height. In comparison, the lower facial height is normal with division 1 malocclusion.

Because the position of the incisors dictates the degree of surgical movement, the orthodontist positions the maxillary and mandibular incisors in the “ideal” position in both the anteroposterior and vertical planes. Failure to adequately decompensate limits the surgical correction and aesthetic outcome. When there is crowding of the lower dental arch, it is frequently necessary to extract the lower first bicusps and retract the anterior dentition. This allows maximal mandibular advancement. In comparison, maxillary crowding in many circumstances can be managed by transpalatal expansion or, when severe, extraction of the upper second bicusps to minimize retraction of the maxillary incisors so as not to limit the mandibular advancement.

Routine leveling of the occlusal plane to obtain arch compatibility needs to be individualized, depending on the severity of the curve of Spee. If the curve of Spee is orthodontically leveled before surgery, this is accomplished by intrusion of the mandibular incisors and will further adversely decrease the lower facial height in patients with class II division 2 malocclusion. It is preferable to skeletally advance the mandible to incisor class I before leveling to maximally increase the lower facial height. The resultant lateral open bite is then closed orthodontically in the postsurgical phase by extrusion of the premolars. With only three-point contact (anterior and the two posterior molar regions), a surgical splint is needed to ensure stability. The greater the lower facial height, the more likely that the leveling should be done by incisor intrusion before surgery. With the exception of the patient with a severe curve of Spee and decreased lower facial height discussed previously, establishing maximal arch compatibility before surgery is important to ensure immediate postoperative stability.

Skeletal advancement is achieved by BSSO of the mandible as described previously. The postsurgical management includes the use of class II elastics to override proprioception and guide the new occlusion in the immediate postoperative period. Orthodontic management then finalizes the coordination of the dental arches by definitive leveling of the curve of Spee, closure of any spaces, and correction of any minor crossbites.



Fig. 27-55 Mandibular deficiency. Mandibular advancement was achieved by BSSO of the ramus.



Fig. 27-56 Mandibular advancement combined with an osseous genioplasty. The patient subsequently underwent a rhinoplasty.

Mandibular Excess (Prognathism)

Patients who have mandibular excess (prognathism) have a concave facial profile where the deformity is primarily isolated to the lower third (Fig. 27-57). However, the vast majority of patients also have some degree of midfacial skeletal deficiency. It is estimated that only 20% to 25% of patients have isolated or “true” prognathism. A number of patients also have overclosure of the mandible, further accentuating the prognathism and midfacial deficiency. Dental examination will show a class III malocclusion with lingually inclined mandibular incisors.

Presurgical orthodontic preparation includes decompensating the mandibular incisors to the “ideal” position; however, in many patients this is limited by the thin alveolar process of the symphyseal region and the thinly attached gingiva. Any transverse width discrepancy must be addressed by orthodontic expansion of the maxillary palate or surgical assistance before formally correcting the class III sagittal discrepancy.

The surgeon's options for correcting the class III skeleton are either the IVO or BSSO of the ramus. The IVO is technically simpler with a lower incidence of inferior alveolar nerve in-



Fig. 27-57 Mandibular excess corrected by BSSO with a setback. In the early recovery phase, the patient is maintained in class III occlusion with the use of dental elastics for neuromuscular adaptation.

jury and is less likely to adversely affect the biomechanics of the temporomandibular joint compared with the BSSO. In addition, when large rotational movement of the mandible is required to correct asymmetry, the IVO is preferred by many on the side that moves posteriorly, whereas on the advancement side, a BSSO is carried out. The disadvantage of the IVO is the prolonged 4- to 6-week period of maxillary-mandibular fixation, because rigid fixation is not possible and it is only suited for setback. However, in most circumstances of class III patients requiring less than 10 mm setback, the BSSO with internal fixation is preferred, because it allows early function. When BSSO setback is carried out, overcorrection by approximately 2 mm should be planned in the surgical splint fabrication because of the postsurgical rebound that occurs as the condyle repositions in the glenoid fossa and the relapsing forces that occur as a result of decreasing the intraoral volumetric space. Class III elastics are required for extended periods with postsurgical orthodontic finalization. For minimal setbacks, instead of a BSSO, it is frequently preferable to correct the class III malocclusion with maxillary advancement from an aesthetic standpoint alone, because the skeletal expansion allows “filling” of the soft tissue envelope. For large setbacks, similarly combining midfacial skeletal advancement will limit the amount of mandibular setback required, and a BSSO rather than an IVO can be accomplished. Present-day aesthetics suggest that the isolated mandibular setback is rarely required.

Sagittal Maxillary Deficiency

Because of the similarity in clinical presentation, maxillary anteroposterior sagittal deficiency has often been diagnosed solely as mandibular excess (prognathism), and a mandibular setback procedure is planned (Figs. 27-58 and 27-59). However, in many circumstances the deformity also includes the midface, and instead of a mandibular setback alone, the patient is better served by a midfacial skeletal advancement either solely or in combination with mandibular surgery. Both have a class III skeletal pattern. Clinically, these patients present with a concave facial profile, deficiency of the maxilla that may extend to involve the zygoma, paranasal deficiency with a narrow alar base, an acute nasolabial angle, a short upper lip, a retrusive upper lip with a thin vermilion, and in many circumstances an accompanying lack of dental display (vertical deficiency). Dental characteristics include a class III molar and canine relationship, maxillary dental crowding with canines blocked out of the dental arches, and in many instances deficient transverse palatal arch width (posterior lingual crossbite), proclined maxillary incisors, and mandibular incisors that are either in the normal position or retroclined.

The orthodontist then eliminates the dental compensations, establishes an ideal incisor position, and coordinates the maxillary and mandibular dental arches. If there is an absolute transverse width discrepancy, this requires palatal expansion as a two-segment LeFort I osteotomy with simultaneous correction of the sagittal discrepancy. The roots of the central incisors then need to be deviated to safely perform the interdental osteotomy. In many patients it is preferable to stage the orthognathic surgery by first correcting the transverse width discrepancy by orthodontic expansion, which may require surgical assistance (surgically assisted rapid palatal expansion), and subsequently correcting the sagittal discrepancy as a single-unit LeFort I procedure to maximize skeletal stability.

With maxillary crowding and the need for incisor retraction, the decision to extract the first or second premolar is based on the extent of crowding and the degree of incisor decompensation needed. When the advancement of the mandibular incisors is limited by lack of attached gingiva and/or minimal alveolar bony support, a mandibular second premolar extraction may be necessary to provide the necessary space. In most circumstances, the maxillary first premolars are extracted, and when needed in both arches, it is combined with extraction of the mandibular second premolars as the most common extraction pattern.

The surgeon then corrects the midfacial skeletal deficiency with a LeFort I osteotomy that is tailored to address the patient's particular deformity. The traditional LeFort I osteotomy is inclined inferiorly from anterior to posterior. Thus when the maxilla is advanced along this incline plane, the sagittal deficiency is corrected. This results in an undesirable decrease in the anterior facial vertical height, and in many cases there is a need to correct the associated vertical deficiency (lack of dental display). The maxillary segment must then be rotated, which results in a significant bony gap and the need for a bone graft. If instead the osteotomy is designed in a reverse plane

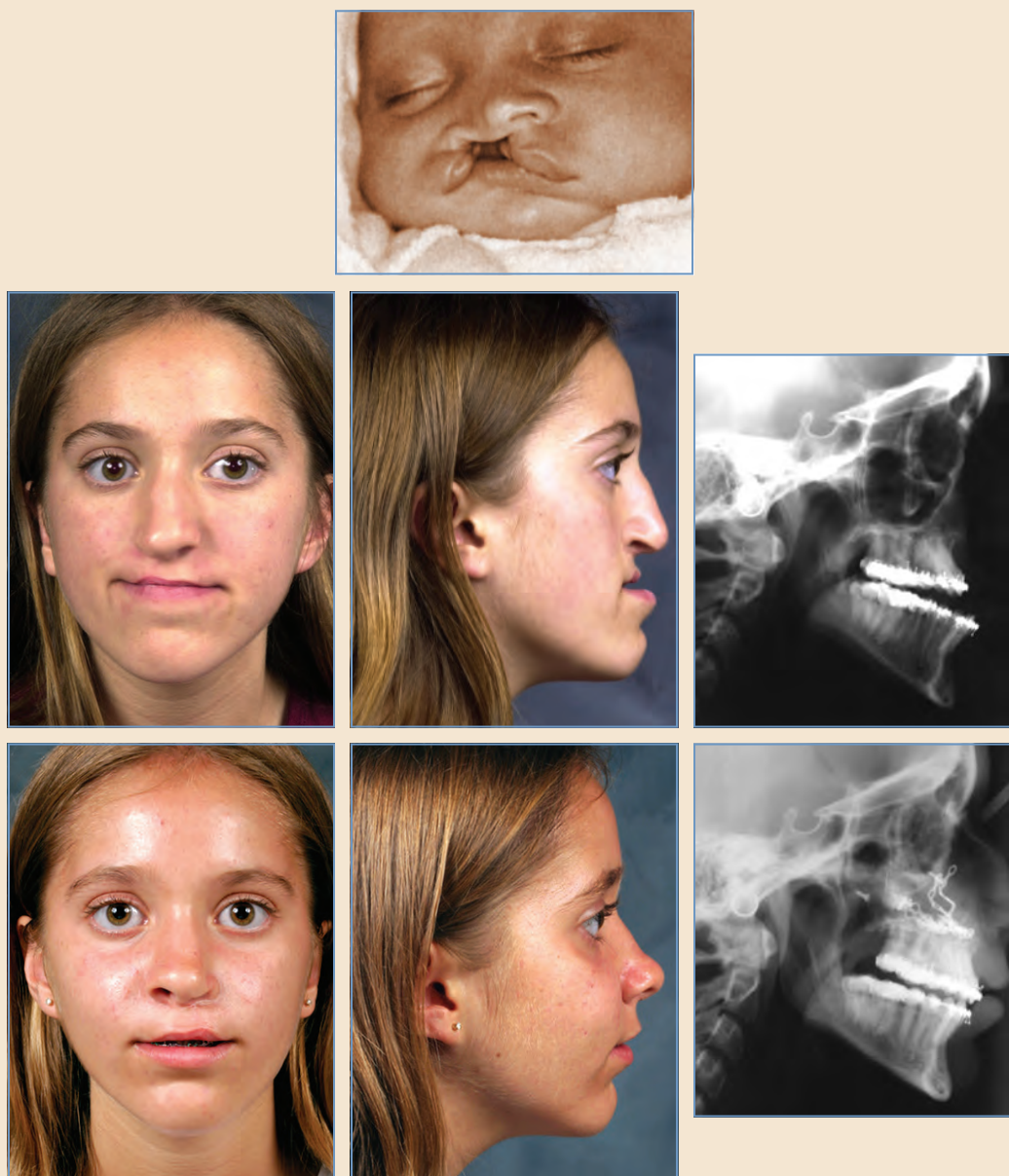


Fig. 27-58 Midfacial skeletal deficiency in a patient with a facial cleft corrected in two stages by reconstructing the cleft alveolar defect with an iliac bone graft followed by a modified LeFort I advancement.

that is inclined superiorly from anterior to posterior, then as the maxilla is advanced to correct the sagittal deficiency, the vertical deficiency is corrected by correspondingly increasing the anterior vertical height. This results in sufficient bony contact without the need for a bone graft in most cases and a stable advancement with titanium plate fixation alone. By varying the inclination of the osteotomy plane, the anterior vertical height can be corrected. When there is severe vertical deficiency, distraction osteogenesis remains the most stable means of achieving significant vertical lengthening. In addition, many patients with maxillary deficiency will also have malar deficiency (flat malar prominence), and the traditional LeFort I osteotomy will not address this, because the osteotomy courses inferior to the ascending lateral zygomatic buttress. The LeFort I osteotomy is then modified to include the body of the zygoma (retruded malar eminence) and, to a varying extent, the zygomatic arch as first described by Obwegeser in 1969 and now known as the “quadrangular” LeFort I osteotomy popularized by Keller and Sather in 1990.^{1,78,84} With these variations, the osteotomy of the posterior lateral maxilla extending superiorly along the posterior aspect of the body of the zygoma is made with some degree of difficulty, and frequently the osteotomy may not be complete and the downfracture may be difficult.



Fig. 27-59 Maxillary deficiency with mandibular overclosure corrected with a modified LeFort I advancement.

Vertical Maxillary Excess

Patients with vertical maxillary excess or long-face syndrome will have an increase in lower facial height and, with clockwise rotation of the mandible, a convex facial profile. The chin is vertically long and retrusive (Figs. 27-60 and 27-61). Bilabial lip incompetence (increased interlabial gap) is present, and mentalis strain occurs with attempted lip closure. There is excessive dental display on repose and a “gummy” smile. The lower lip appears protrusive without support of the upper dentition. The alar base is frequently narrow, the paranasal regions are deficient, and there is lack of malar projection in many patients. Intraoral examination reveals an anterior open-bite deformity in most of these patients, a high-arched palate with a V-shaped transversely narrow maxilla, and dentition in palatal crossbite. Although the occlusal relationship is class II in most cases, class I and class III can occur with vertical maxillary excess. Downward and posterior rotation of the mandible (clockwise rotation of the mandibular plane) resulting from excessive vertical maxillary growth will make the associated mandibular deficiency appear worse and the mandibular excess not as severe. Although an anterior open bite is the usual presentation, in patients with vertical maxillary excess and a deep bite, the lower facial height may not appear to be increased, and excessive dental display is camouflaged by the lower lip.

Orthodontic management involves elimination of dental compensations and leveling of the mandibular arch. In patients in whom the open bite is severe and there is an excessive reverse curve in the maxillary arch, it is preferable to level the arch in segments with root deviations to allow interdental osteotomies and surgical leveling with a three-segment LeFort I procedure. When the vertical discrepancies are minimal, the arch can be leveled in a single plane, and the open bite can be corrected by a differential posteroanterior impaction single-unit LeFort I procedure. The open bite should not be closed orthodontically to minimize recurrence. Relief of dental crowding and transverse palatal expansion (surgically assisted) should be addressed before definitive orthognathic surgery.



Fig. 27-60 This patient with vertical maxillary excess underwent presurgical orthodontic treatment. The transverse maxillary deficiency was corrected in the first stage through transpalatal expansion.

In most patients, correction of vertical maxillary excess requires double jaw surgery: differential LeFort I impaction of the maxilla either as a single segment or multisegment with mandibular BSSO advancement and a genioplasty. It is rare that LeFort I impaction, with or without a genioplasty with reliance on autorotation of the mandible, will result in a stable occlusion and satisfactory aesthetic outcome. Prediction cephalometric tracings are beneficial to assess the need for concomitant mandibular surgery. Although preoperative planning will approximate the amount of bone that needs to be resected to achieve the desired dental display based on clinical examination and desired final aesthetics, en bloc removal of the predetermined amount will likely leave large gaps between bony surfaces with superior repositioning of the maxilla. To avoid this, less bone should be resected initially and the dental display should then be adjusted intraoperatively by selectively and sequentially removing bone from points of bony interferences with the use of rongeurs as the maxillary-mandibular complex with an intermediate splint is rotated counter-clockwise. This will frequently result in slotted joints that lock the mobilized maxilla into its base similar to “Lincoln logs.” Failure to remove adequate bone from the posterior maxilla and medial maxillary walls will prevent unrestrained superior repositioning of the maxilla, and frequently the condyle is brought out of the fossa with rotation of the complex. This will become evident when the maxillary-mandibular fixation is released and the mandible fails to rotate appropriately into

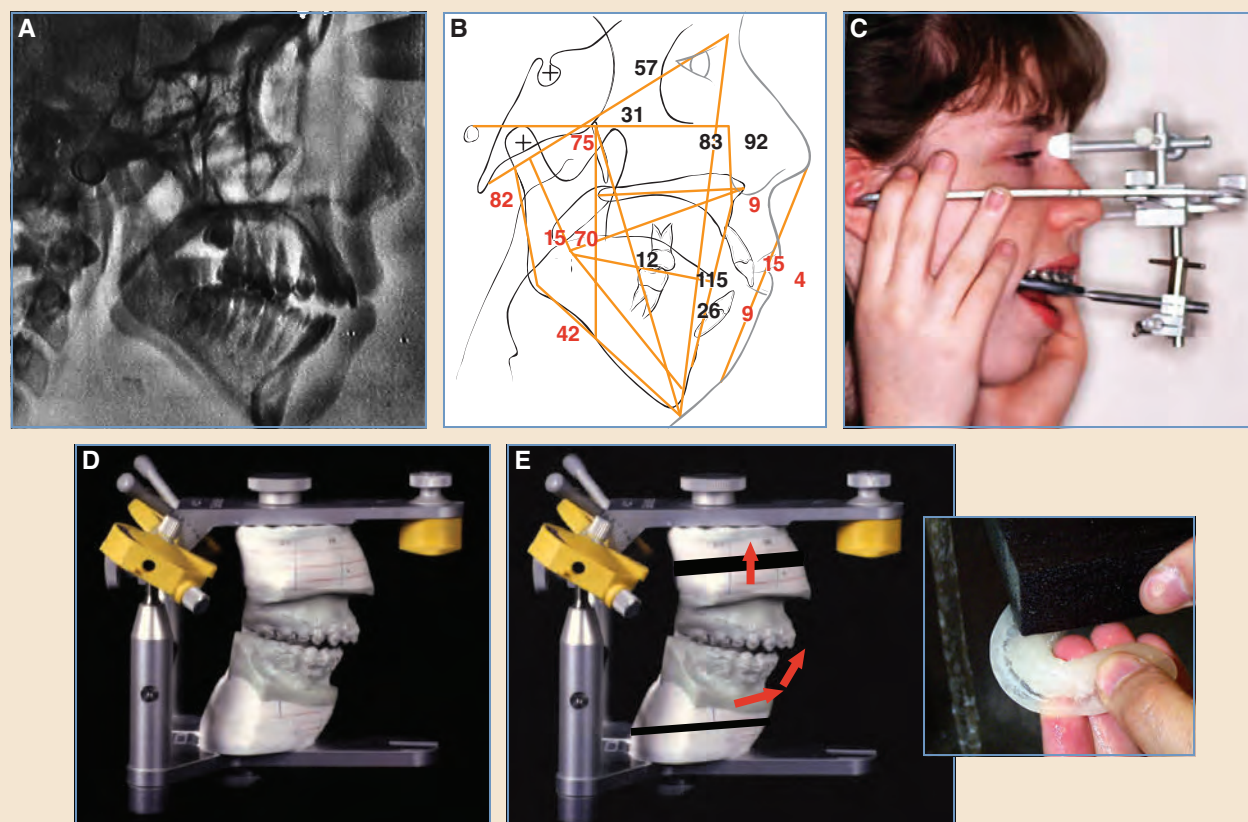


Fig. 27-61 Presurgical planning involving cephalometric analysis (A and B), facebow transfer (C), and model surgery (D and E).

Continued



Fig. 27-61, cont'd F, Intraoperative view showing resection of the maxilla for vertical impaction. G-J, Postoperative outcome with finalization of orthodontic occlusion.

the intermediate splint. In addition, with impaction of the maxilla, the base of the septum and vomer should be trimmed, and inferior turbinectomies may need to be carried out to minimize significant septal buckling and airway compromise from decreased nasal cross-sectional area.

Undesirable changes can occur with maxillary impaction that alter the alar base width, the columellar-labial angle, nasal tip projection, and lip contour. These must be assessed before surgery and addressed intraoperatively with alar cinch sutures and contouring the nasal piriform to adjust the alar vestibule, V-Y closure of the lip to alter the lip contour, and an osteotomy that repositions or contours the anterior nasal spine to control the nasal labial angle and upper lip length. Soft tissue alterations are not necessarily always predictable and must be discussed with the patient because secondary surgery involving the lip and nose may be needed to achieve the patient's aesthetic goals.

Facial Skeletal Asymmetry

Patients may also have facial skeletal asymmetry as a result of congenital deformities (hemifacial microsomia), developmental problems (unilateral condylar hyperplasia), or trauma to the condylar region (Fig. 27-62). The principles outlined earlier apply, although correction of such dentofacial deformities is typically more complex, because there is asymmetry in all three planes. Model surgery must be accurate, and CBCT scans will often be needed to guide the surgical planning. In addition to the skeletal asymmetry, many patients will have soft tissue components that require correction after the skeletal framework is in a more symmetrical position.

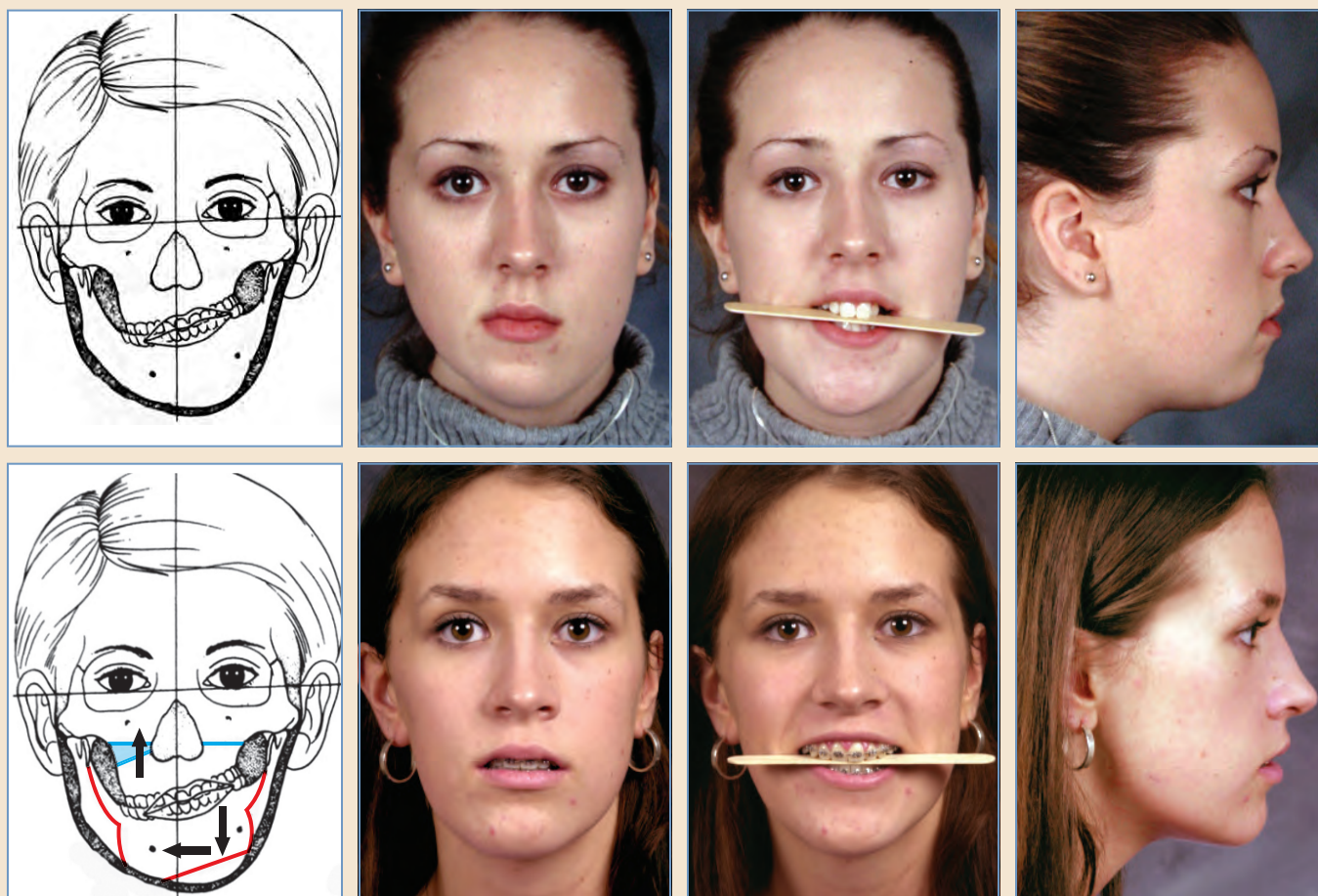


Fig. 27-62 Patient with facial skeletal asymmetry (unilateral left condylar hyperplasia) corrected by asymmetrical LeFort I osteotomy, BSSO, and genioplasty.

MANAGEMENT OF CLEFT-RELATED DENTOFACIAL DEFORMITIES

Orthognathic surgery to correct dentofacial deformities in patients with facial clefts is a more complex undertaking, because such patients present with not only skeletal considerations, but the soft tissue component also plays a significant role in the management and outcome. Although the goal of orthognathic surgery in all patients is to achieve ideal occlusion, in patients with clefts, this often requires performing multiple-segment osteotomies and overcoming the restricting forces of the soft tissue–scar tissue impediment to maxillary advancement. Furthermore, the surgeon must take into account poor bone stock. All these factors must be carefully considered and accurately evaluated during the preoperative planning stages. A comprehensive strategy that addresses each of these variables is of paramount importance for achieving an excellent result.

Approximately 25% to 30% of patients who have undergone surgical correction of cleft lip/palate develop midface retrusion severe enough to require orthognathic correction. Maxillary hypoplasia resulting in class III malocclusion is the typical deformity seen in this patient population. The cause of maxillary hypoplasia is twofold. Evidence shows that inelastic scar tissue from the original palatoplasty restricts facial growth and suggests an intrinsic midfacial growth deficiency unrelated to surgery (that is, it is an inherent component of the congenital anomaly).⁸⁵ Although mild class III malocclusions may be treated with orthodontics alone, orthognathic surgery is needed to ideally correct a more severe class III dentofacial deformity.

Preoperative Considerations

The chance of a favorable surgical outcome is optimized if presurgical planning is performed in conjunction with a cleft/craniofacial team. Speech pathologists play an integral role in evaluating the velopharyngeal mechanism and the potential effects that maxillary advancement may have on speech nasality and articulation. Preoperative videonasoscopy has been shown to yield information that can aid in predicting postoperative hypernasality.

Patients with a cleft lip and/or palate anomaly have several anatomic differences compared with unaffected patients. The maxilla is typically deficient in both the anteroposterior and vertical dimensions. Because midface retrusion can be significant, it frequently appears that the mandible is prognathic, but true mandibular prognathia is rare. The apparent prognathia is relative and a result of the maxillary deficiency. Also, because of lesser segment collapse, the dental midline is often deviated toward the cleft side, and this must be taken into consideration for proper midline repositioning.

Despite having undergone alveolar bone grafting, many of these patients have deficient or missing bone in the region of the alveolus. Persistent palatal fistulas may also be present. The lateral incisor is frequently missing in these patients, and closure of this space must be taken into account at the time of treatment planning. Various options for prosthetic reconstruction of this edentulous space are described in Chapter 29. If a large fistula is present in the alveolus, modifications of the LeFort I procedure can be performed to facilitate a tension-free alveolar closure. These are discussed in the following section.

Surgical Aspects

Orthognathic surgery for patients with cleft lip/palate is similar to that for patients without clefts, except for several important modifications necessary to maintain blood supply and assist in associated primary palatal alveolar residual cleft oronasolabial fistula closure.

In patients with unilateral cleft lip, the standard incision can be made with little jeopardy to the premaxillary blood supply (Fig. 27-63, *A*). Each side of the cleft requires an incision made similar to that of an alveolar bone graft incision (Fig. 27-63, *B*). This allows two-layer closure of the palatal and nasal mucosa. If supplemental bone grafting is required at this time, harvested bone can be placed into the alveolar gap after fixation has been applied. If a wide fistula is present, the maxillary segments can be compressed to reduce the size of the alveolar space (Fig. 27-63, *C*). This maneuver ensures that the soft tissue closure is under minimal tension, and the chance of fistula closure is optimized. Compressing the maxillary segments or congenital absence of the lateral incisor can place the canine adjacent to the central incisor. Although this may not be a great aesthetic concern, a restorative dentist can fabricate a prosthetic crown for the canine to make it look like a lateral incisor.

With patients who have bilateral clefts, care must be taken to avoid making the vestibular incision across the premaxilla. The premaxillary blood supply is from the vomer and buccal mucosa. Because the vomer will be split, most of the blood flow to the premaxilla will be from the premaxillary buccal mucosa. A circumvestibular incision that violates this mucosa can severely

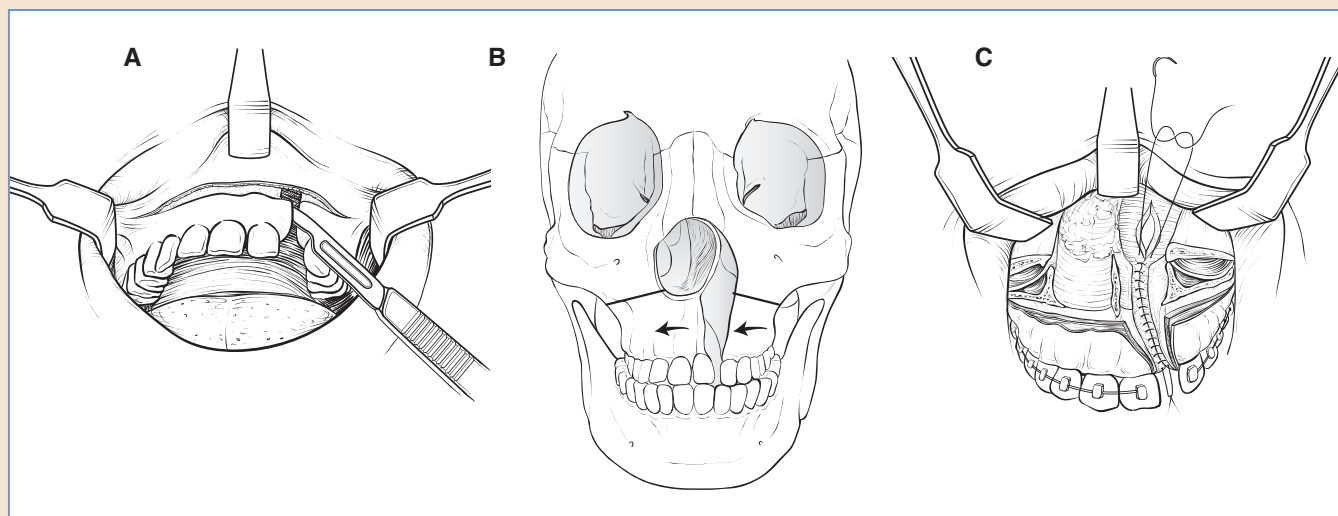


Fig. 27-63 *A*, For patients with a unilateral cleft lip, the incision is made similar to a standard LeFort I osteotomy, except an alveolar dissection is used if supplemental bone grafting or fistula closure is necessary. *B*, The LeFort osteotomy allows compression of the maxillary segments if necessary to close a preexisting fistula. *C*, Fistula repairs are easier after compression of the segments and exposure of nasal and palatal tissue.

jeopardize the blood supply of the premaxillary segment. To minimize the risk of complications, the incision is stopped just lateral to the alveolar cleft on each side (Fig. 27-64, *A*). Reflection of the mucosa from the premaxilla is minimized to preserve the blood supply. The osteotomy of the premaxillary segment is made from a posterior approach just anterior to the incisive foramen to allow mobilization of the segment without violation of the buccal mucosa (Fig. 27-64, *B*). Similar to a unilateral cleft maxilla, residual fistulas and inadequate alveolar bone may be present. If either is identified, it can be corrected by a two-layer mucosal closure and bone grafting into the alveolar defect (Fig. 27-64, *C*). If large gaps are present that may jeopardize fistula closure, the segments can be compressed at the alveolar gaps to reduce the tension of the repair (Fig. 27-64, *D*). Postoperative orthodontics and prosthetic restorations of the teeth can correct most postoperative dental aesthetic irregularities.

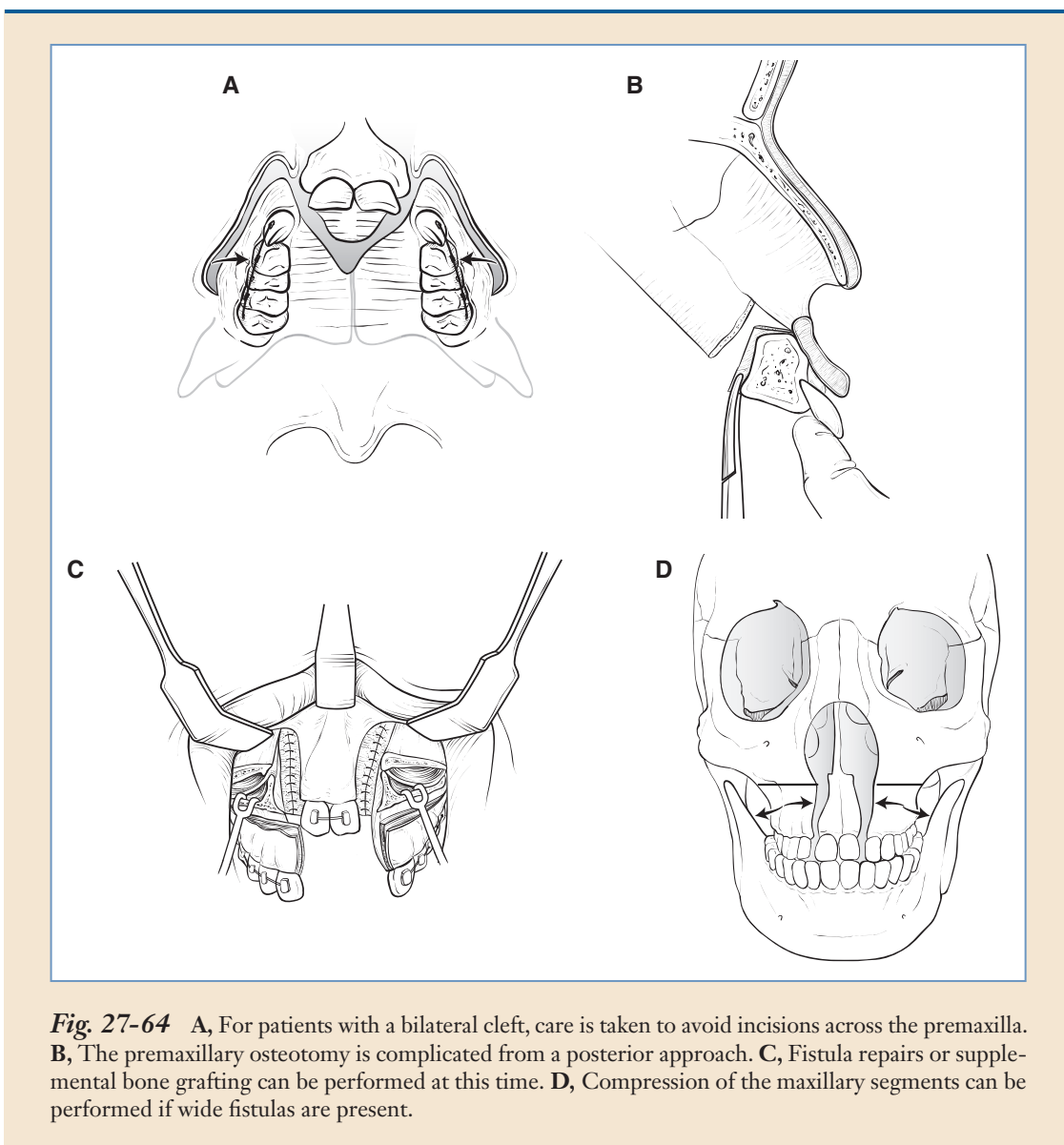


Fig. 27-64 *A*, For patients with a bilateral cleft, care is taken to avoid incisions across the premaxilla. *B*, The premaxillary osteotomy is complicated from a posterior approach. *C*, Fistula repairs or supplemental bone grafting can be performed at this time. *D*, Compression of the maxillary segments can be performed if wide fistulas are present.

Velopharyngeal Dysfunction and Speech

It is generally accepted that velopharyngeal dysfunction (VPD) in patients with cleft lip/palate is caused by a malalignment or shortening of the palatal musculature, skeletal growth and development, and/or surgical sequelae that can lead to abnormal structural relationships. Alveolar fistulas and a constricted arch can contribute to resonance disorders (hypernasality or hyponasality) or articulation defects (errors in formation of consonants). Given the intricate attachment of the maxilla to the muscular apparatus of the velum, it follows that moving the maxilla can change velopharyngeal function.

Janulewicz et al⁸⁶ performed a retrospective study of the change in velopharyngeal function in cleft lip/palate patients who underwent maxillary advancement with or without a mandibular setback procedure over a 21-year period. As summarized in Table 27-3, their study showed a decline in competent velopharyngeal function (42% to 18%) and an increase in both borderline insufficiency (9% to 22%) and complete VPD (13% to 20%). The authors also noted that the quality of speech declined, as evidenced by the increase in overall objective speech score from 2.46 to 4.24 (the higher the score, the worse the speech). In contrast, the authors noted that articulation defects improved, although the improvements did not achieve statistical significance. Preoperatively, 84% (46 patients) had at least one articulation defect compared with 73% (40 patients) postoperatively.

Other studies have shown similar results or no change in VPD function. Both Watzke et al⁸⁷ and more recently Phillips et al⁸⁸ have shown that the extent of anteroposterior maxilla movement is unrelated to velopharyngeal functional deterioration and is not a useful predictor. In a study of 25 patients (16 unilateral complete and 9 bilateral complete cleft lips/palates), the authors showed that all patients who had perceived hypernasal speech before their surgery had hypernasality after advancement. Furthermore, 9 of 12 patients (75%) who had preoperative

Table 27-3 Comparison of Preoperative and Postoperative Speech Variables

Total Number of Patients: 54	Preoperative Evaluation Percentage (n)	Postoperative Evaluation Percentage (n)
VP function: competent	42 (23)	18 (10)
VP function: borderline competent	36 (20)	40 (22)
VP function: borderline incompetent	9 (5)	22 (12)
VP function: complete VPD	13 (7)	20 (11)
Normal nasality	40 (22)	40 (22)
Mild hypernasality	18 (10)	29 (16)
Moderate hypernasality	4 (2)	15 (8)
Severe hypernasality	4 (2)	2 (1)
Hyponasality	33 (18)	15 (8)
Reduced sibilant IOAP	26 (14)	35 (19)
Reduced fricative IOAP	16 (9)	26 (14)
Reduced plosive IOAP	6 (3)	22 (12)
Anterior dentition errors	64 (35)	47 (26)
Mean speech score	2.46	4.24

IOAP, Intraoral air pressure; VP, velopharyngeal; VPD, velopharyngeal dysfunction.

nasopharyngoscopy showing borderline or inadequate closure developed VPD postoperatively. Furthermore, they found that nasoendoscopy adds little to perceived speech testing in its ability to predict which patients develop VPD after maxillary advancement. Based on these results, the researchers concluded that preoperative assessment can predict postoperative speech and velopharyngeal function and that nasoendoscopy plays a minor role in the preoperative evaluation of patients without fistulas and borderline or hypernasal speech.

Because elements of speech are linked to hard and soft tissue relationships, advancing the maxilla would seemingly affect articulation; however, data from multiple studies appear to be inconsistent. Witzel and Munro⁸⁹ showed that improving skeletal malocclusions with orthognathic surgery improved articulation, especially for the production of sibilant sounds. In contrast, Dalston and Vig⁹⁰ found no improvement in articulation after surgery.

Thus it appears that although a positive effect on articulation might be achieved by orthognathic surgery, it might be at the expense of velopharyngeal function. Because there is uncertainty about how much maxillary advancement could affect speech, the physician should discuss possible changes in speech with the patient and family before surgery.

Skeletal Stability and Relapse With Conventional Cleft Orthognathic Surgery

Posnick⁹¹ performed a retrospective study evaluating relapse in patients who had undergone orthognathic surgery for cleft lip/palate between 1987 and 1990. The author found that there was no significant difference in outcome between patients who had maxillary surgery alone and those who had surgery on both jaws. Furthermore, the outcome did not vary significantly with the type of autologous bone graft used or the segmentalization of the osteotomy.

All 35 patients included in the study had a modified LeFort I maxillary osteotomy with varied degrees of horizontal advancement, transverse arch widening, and vertical change. Eleven of the 35 patients (32%) also required mandibular surgery consisting of BSSO.⁹¹ In 13 of 35 patients (37%), a pharyngoplasty was in place at the time of the LeFort I osteotomy.⁹¹ The mean horizontal advancement achieved for the group was 6.9 ± 2.6 mm at 1 week; 5.3 ± 2.7 mm was maintained 1 year later (mean relapse 1.6 mm). In 11 of 35 patients (32%), the relapse was less than 1.0 mm. For the 13 patients (37%) who had pharyngoplasty at the time of the LeFort I osteotomy, the mean horizontal advancement was 8.2 mm immediately after surgery and 6.5 mm 1 year later. The stability of the vertical displacement was also evaluated. No maxillary vertical change was necessary in 12 of 35 patients (34%). The mean vertical displacement of the maxilla in patients who underwent vertical repositioning was 2.1 ± 2.4 mm at 1 week; 1.7 ± 2.0 mm was maintained 1 year later. The author concluded that neither horizontal nor vertical relapse was related to the extent of movement. The overjet seen on cephalometric radiographs 1 year postoperatively was maintained in all patients, whereas a positive overbite was maintained in only 30 of 35 patients (85%).⁹¹

Other investigators have found a correlation between relapse and the degree of advancement.⁹² To identify factors associated with relapse after orthognathic surgery in patients with cleft lip/palate, Hirano and Suzuki⁹³ performed a retrospective study on 58 patients with cleft lip/palate who underwent orthognathic surgery over a 10-year period. They identified the following factors related to relapse:

1. Horizontal advancement. The mean horizontal relapse was 24.1% of the mean advancement. There was significant correlation between extent of relapse and advancement. The authors report that complete surgical mobilization of the maxilla is important for preventing relapse.

2. Vertical displacement inferior positioning. The mean inferior vertical elongation was 3 mm, with a relapse of 2.1 mm. The authors recommended a 2 mm overcorrection with inferior positioning of the maxilla.
3. Rotation, clockwise or counterclockwise. Most surgical rotation was lost, and relapse was seen with both clockwise and counterclockwise rotations. The authors recommended overcorrection to mitigate the effects of relapse.
4. Type of cleft. Orthognathic surgery for a bilateral cleft was more likely to result in relapse. The authors attributed this greater likelihood to increased scarring of palatal tissues and multiple missing teeth.
5. Previous alveolar bone grafting. Although studies have reported the value of alveolar bone grafting for establishing advancement stability and minimizing relapse, Hirano and Suzuki⁹³ found no association between alveolar bone grafting and the rate of relapse in patients with unilateral cleft lip/palate.
6. Number of missing teeth. No correlation was found between the number of missing teeth and relapse, although the authors stressed that multiple missing teeth can compromise the stability of the occlusion.
7. Type of orthognathic surgery. There was no difference in the relapse rate between patients who underwent maxillary surgery alone and those who underwent two-jaw surgery.

Role of Distractional Osteogenesis in Midfacial Advancement

Patients with mild-to-moderate maxillary hypoplasia are good candidates for traditional or conventional orthognathic surgery; however, those with severe maxillary retrusion (greater than 10 mm) are prone to a significant amount of relapse. Several studies have shown that patients with severe maxillary deficiency have less predictable results and a higher rate of relapse.⁸⁻¹² Furthermore, the need for revision and prolonged orthodontics is higher. This subset of patients might benefit from advancement by distraction osteogenesis.

Distraction osteogenesis at the LeFort I level has been recommended for the early treatment of severe midface deficiency for patients in the mixed dentition stage. Cho and Kyung⁹⁴ conducted a retrospective study to evaluate the long-term stability of maxillary distraction osteogenesis with a rigid external device, based on a 7-year experience. In their study, the mean distraction length was 13.6 mm immediately after distraction (Table 27-4). However, at the 6-month follow-up, the mean distraction length was 10.8 mm and 10.4 mm at the 1- to 6-year follow-up. Based on their results, the mean postoperative relapse rate was 23% at 1 to 6 years. Preoperatively, the mean SNA angle was 78.5 degrees, and immediately after distraction it was 90.7 degrees. Six months postoperatively, the mean SNA angle was 87.2 degrees and 86.9 degrees 1 to 5 years postoperatively. These results show that most relapse occurs in the first 6 months. The relapse rate increased

Table 27-4 External Distraction Outcomes

	Immediate Postoperative Outcome	6 Months Postoperatively	1 to 6 years Postoperatively
Mean length	13.6 mm	10.8 mm	10.4 mm
Mean relapse rate	N/A	20.7%	23.0%

up to 6 months after distraction and was not statistically significant between 1 year and 6 years after distraction. They concluded that a 20% to 30% overcorrection is required with maxillary advancement to compensate for the relapse.

Polley and Figueroa⁹⁵ studied 16 patients between 5 and 25 years of age who underwent correction of maxillary hypoplasia with a rigid external distraction device. The patients underwent distraction followed by a 2- to 3-week rigid retention phase and achieved a mean advancement of 11.7 mm. The authors reported no relapse at the 4- and 12-month follow-up intervals. In later reports they attributed decreased facial convexity at the 1-year follow-up to a combination of maxillary resorption, forward mandibular growth, and closed rotation of the mandible.

The benefits of rigid external distraction are the versatility and flexibility of both the amount and direction of the distraction process. With a skull-anchored, external headframe distractor attached to an orthodontic splint, precise control of the maxillary segments can be achieved with accurate, consistent, and unlimited distraction. However, the only aspect of this system that is rigid is the halo, which attaches to the head. The maxilla is not rigidly held in its postoperative position after advancement. Patients are allowed to function with an osteotomized maxilla. Although studies have not shown a higher incidence of nonunion with this concept, it contradicts principles of fracture management.

Kumar et al⁹⁶ compared the correction of severe maxillary retrusion using distraction via internal devices with conventional orthognathic surgery. Fifty-one patients with cleft lip/palate were divided into three groups based on the extent of maxillary retrusion and type of orthognathic treatment: group 1 (n = 20): mild-to-moderate deficiency (less than 10 mm) treated with a conventional orthognathic procedure; group 2 (n = 11): severe deficiency (10 mm or more) treated with conventional orthognathic surgery; and group 3 (n = 20): severe deficiency (10 mm or more) treated with distraction osteogenesis.

The results showed that group 1 patients had a mean advancement of 7 mm (Table 27-5). In this group, all patients finished with a class I occlusion and had no complications, relapse, or revisions. Group 2 patients had a mean horizontal advancement of 11 mm. In this group, one patient (9%) had a class III malocclusion postoperatively, and six of 11 patients (55%) had class III malocclusion at follow-up. Seven of 11 (64%) developed relapse, and five of 11 (45%) underwent reoperation. Group 3 patients had a mean advancement of 25 mm. At follow-up, a 15% relapse rate was observed (3 of 20 patients).

Although group 2 patients underwent less advancement than group 3 patients, group 2 patients had a higher relapse rate. The authors suggested that the gradual skeletal advancement offered by distraction provides greater advancement with less relapse in patients with severe maxillary deficiency. When conventional orthognathic surgery was used for group 2 patients, the lack of gradual expansion affected the surrounding soft tissue scars, which most likely contributed to the high relapse rates seen in this group. The authors also suggested that gradual advancement with distraction osteogenesis allows the LeFort I segment to move a greater distance and remain stable in its new location because of slow scar tissue adaptation.

The effects on speech were compared between distraction and orthognathic procedures. Patients who underwent distraction had better speech scores and less VPD than acutely advanced patients. Furthermore, nasal endoscopy in selected patients showed that those who underwent distraction had fewer structural problems with oronasal closure than patients in the orthognathic group. This study indicates that for patients with severe maxillary deficiency, LeFort I distraction may offer better results than traditional orthognathic surgery.

Orthognathic surgery is typically performed after skeletal growth is complete so that the patient's mandible does not outgrow the surgical correction achieved by maxillary advancement. Delaying the procedure also avoids the risk of injuring tooth buds or roots of the permanent dentition, if the teeth have not erupted beyond the site of the planned osteotomy. The benefit of

the traditional orthognathic surgical procedure is precise control of the occlusion and application of rigid fixation to secure the maxilla's position.

Unlike the one-stage orthognathic procedure, advancement by distraction requires at least two operative procedures, and the patient must also endure the appliance for 8 to 12 weeks. The LeFort I osteotomy for distraction is essentially the same as that performed for traditional orthognathic surgery, and thus the risk of injury to tooth buds and the potential to outgrow the correction are the same. Also, if distraction is performed at an earlier age, the risks are higher than with traditional orthognathic surgery. Final control of occlusion is not as precise with internal distraction, and mobility is present after external distraction. Surgical distraction must offer clear benefits over traditional orthognathic surgery to be considered for treatment. Indications for the use of maxillary distraction over traditional orthognathic surgery are reduced rates of relapse and VPD. With an extremely deficient maxilla for which the surgeon is not comfortable with his or her ability to achieve adequate advancement with orthognathic techniques, distraction allows a slow adaptation of the scarred palatal tissues, possibly reducing relapse. In addition, distraction has been shown to have a lower postoperative incidence of VPD.^{95,96}

Unless distraction achieves perfect occlusion without relapse and mandibular overgrowth, the patient will still require subsequent orthognathic surgery. In this situation the question arises regarding whether the long process of distraction osteogenesis followed by an orthognathic procedure offers any advantages over two-stage LeFort I osteotomies. We think that although distraction osteogenesis is a valuable tool, its indications must be carefully weighed against those of traditional orthognathic techniques.

Table 27-5 Cleft Orthognathic Outcome

	Group I: Mild-to-Moderate Deficiency (mm)	Group II: Severe Deficiency (mm)	Group III: Severe Deficiency (mm)
Deficiency	≤10	≥10	≥10
Number of patients	20	11	20
Type of procedure	Orthognathic surgery	Orthognathic surgery	Internal distraction
Mean advancement	7	11	25
ΔX preoperative to postoperative	5	7.22	16.5
ΔX postoperative to follow-up	4.9	5.8	16.1
ΔY preoperative to postoperative	−0.5	−0.4	−1.1
ΔY postoperative to follow-up	−0.5	−0.2	−1.0
Other Data			
Number of relapse patients	0	7	3
Relapse rate	0%	64% (7/11)	15% (3/20)

ΔX *Postoperative to follow-up*, Horizontal change from postoperative to follow-up; ΔX *preoperative to postoperative*, horizontal change from preoperative to postoperative; ΔY *postoperative to follow-up*, vertical change from postoperative to follow-up; ΔY *preoperative to postoperative*, vertical change from preoperative to postoperative.

COMPLICATIONS OF ORTHOGNATHIC SURGERY

Although complications can occur during any of the dental, orthodontic, or surgical phases of treatment, complications specifically related to the surgical procedures are summarized in Box 27-1. The specific problematic issues related to cleft orthognathic surgery were discussed in the preceding section. The complications discussed in this section are applicable to all patients undergoing orthognathic surgery.

Airway

Airway management is critical because nasoendotracheal intubation is necessary to facilitate intraoperative maxillary-mandibular fixation. Nasal intubation can be difficult in patients with clefts requiring orthognathic surgery who have previously undergone pharyngeal flap surgery. Intubation can be accomplished with fiberoptic guidance or exchanged for a stent after oral intubation. Occasionally, when nasal intubation is not possible, the oral tube can be brought out retromolar at the time of maxillary-mandibular fixation, although this is less than ideal, and unplanned extubation can occur. During the LeFort I surgical procedure, the nasotracheal tube is particularly at risk of septal-vomerine disjunction and the osteotomy of the lateral nasal wall. The nasotracheal tube can be lacerated or severed either by the osteotome or by the reciprocating saw.⁹⁷⁻⁹⁹ An immediate air leak or loss of air exchange is evident when blood is within the endotracheal tube. It is preferable to perform the septal disjunction first with a guarded osteotome to facilitate placement of the reciprocating saw as the septum and nasotracheal deflects to the opposite side and minimizes the risk of injury to the tube. The maxillary osteotomy with a reciprocating saw should be directed from medial to lateral in the opposite direction of the nasotracheal tube. If

Box 27-1 Complications of Orthognathic Surgery

LeFort I Procedure

- Arteriovenous fistulas (carotid-cavernous sinus)
- Avascular necrosis
- Infraorbital nerve traction injury
- Injury to the internal maxillary artery and its branches
- Injury to Stensen's duct
- Malocclusion
- Maxillary sinusitis
- Nasal septal deviation and buckling
- Nasopalatine nerve transection (inevitable)
- Ophthalmic and lacrimal duct injury
- Unanticipated fractures (pterygoid plate, sphenoid bone, middle cranial fossa)

VPD Osseous Genioplasty

- Inferior mandibular border contour irregularity
- Gingival recession
- Mental nerve injury
- Ptosis of the mentalis

BSSO

- Avascular necrosis
- Bleeding (inferior alveolar artery, masseteric artery)
- Condylar resorption
- Inferior alveolar nerve injury
- Malocclusion
- Malpositioned proximal segment
- Unanticipated fractures

Common to All Procedures

- Devitalization of teeth
- Hardware issues
- Injury to teeth
- Malunion/nonunion
- Postoperative infection
- Relapse
- Unanticipated fractures

injury occurs, exchange of the nasotracheal tube is easier before the maxillary osteotomy is completed.¹⁰⁰ The vast majority of patients are extubated immediately after the surgical procedure.¹⁰¹ Internal fixation with titanium plates and screws and postoperative maxillary-mandibular fixation with dental elastics over wire fixation have reduced the likelihood of prolonged postoperative intubation.¹⁰²⁻¹⁰⁹ Intermaxillary fixation increases airway resistance, reducing peak air flow, and patients who have undergone a LeFort I procedure also will, of necessity, have nasal airway obstruction.^{104,105} For these reasons, many prefer no maxillary-mandibular fixation or only guiding elastics in the immediate recovery period.

Hemorrhage

The use of local anesthesia with a vasoconstrictor and controlled hypotensive anesthesia will significantly reduce generalized bleeding and the need for blood transfusion while greatly improving the view of the operative field.^{106,107} Significant hemorrhage is uncommon; when it occurs, it is more likely with maxillary osteotomies.^{102,108} With maxillary osteotomy, the vessels at risk include the greater palatine vessels, maxillary artery, and pterygoid plexus.^{81,109} Bleeding from the descending palatine vessels occurs when the posterior aspect of the lateral nasal wall osteotomy is made, and in most circumstances the bleeding spontaneously stops. When bleeding persists, it is easily controlled with vascular clips or bipolar electrocautery after the maxilla is downfractured.

Because the separation of the pterygopalatine plate with an osteotome is not directly visible, placement of the osteotome at the inferior aspect by palpation is critical to avoid injury to the vessels within the pterygopalatine fossa.¹¹⁰ The margin of safety is approximately 10 mm.⁹⁸ Hemorrhage, when it occurs, can be rapid and is usually controlled by densely packing the fossa with cottonoid. In the vast majority of patients, bleeding, although it may be significant, is easily controlled with localized measures. Direct ligation of the maxillary or carotid artery is necessary only in rare circumstances.¹¹¹ In general, bleeding persists until the maxilla is downfractured and will nearly always be significantly diminished by the time the maxilla is repositioned with internal fixation.

In contrast, the incidence of significant hemorrhage with mandibular osteotomies (BSSO or IVO) is extremely low.¹¹²⁻¹¹⁶ The vessels at risk include the inferior alveolar artery, maxillary artery, facial artery, retromandibular vein, and pterygoid venous plexus.¹¹⁴ With subperiosteal dissection and the use of a medial ramus retractor, the inferior border retractor and lateral splitting will avoid vascular injury in most cases. With loss of the subperiosteal dissection plane (supraperiosteal), especially when exposing the medial surface of the ramus, injury to the vessels occurs, making the medial ramus osteotomy difficult. The use of cottonoid to pack the medial ramus will usually stop the bleeding, and in rare instances the split may need to be completed before the bleeding from the posterior ramal border can be controlled.

Despite all precautions, uncommon vascular complications that may be life-threatening can occur. These include arteriovenous fistulas, false aneurysms, carotid cavernous fistulas, and carotid artery thrombosis.¹¹⁷⁻¹²⁰

Vascular Compromise

With degloving, osteotomy, and repositioning of the facial skeletal elements, there is a significant reduction in the blood supply. The vascular compromise affects not only the skeletal component but also the dentition and associated soft tissue components (pulp, periodontium, and gingiva). In the vast majority of cases, vascular compromise is transient and has no significant clinical impact on the outcome.^{121,122} The degree of vascular compromise depends on the placement of the incision, the extent of degloving of the soft tissue, and the degree of mobilization and movement

of the segment. Additional factors that compromise the vascularity may include previous surgery for cleft reconstruction and adjuvant radiation therapy for head and neck cancer.

The vascularity of the LeFort I downfractured segment is derived primarily from the soft palate and buccal soft tissue pedicle, because the greater palatine vessels are frequently divided during the osteotomy and mobilization.^{80,81,110-124} Although the reduction in the blood supply is transient, devitalization of the teeth, periodontal defects, and segmental bone loss have been described.^{125,126} Many of these problems have been attributed to incisions that compromise the vascularity, excessive stripping of the periosteum, compromised palatal mucosa (previous cleft palate surgery), interdental osteotomies with loss of the attached gingiva, and transverse expansion with excessive stripping of the palatal mucosa. When intraoperative cyanosis of the gingiva is noted and the vascularity does not return with reversal of the hypotension, the surgeon should consider returning the maxilla to its original position and reassessing the surgical approach. When this occurs, readvancement can be gradual with the use of distraction osteogenesis, either surgically controlled with implanted devices or orthopedic elastic traction forces.

As with maxillary surgery, avascular necrosis with mandibular surgery is uncommon.^{121,127} The blood supply to the mandible is derived from the inferior alveolar artery and primarily from the nutrient vessels by way of the muscular attachments of the masseter and pterygoid muscles.¹²⁸ With the sagittal split ramal osteotomy, extensive stripping of the masseter from the buccal cortex may compromise the blood supply to the distalmost region of the proximal segment where internal fixation hardware is placed. Avascular necrosis, although uncommon, can occur, with resulting loss of fixation and skeletal stability. The blood supply to the distal element is derived from the attachment of the pterygoid muscle to the lingual cortex. Division of the pterygomasseteric sling, which is needed to allow adequate mobilization, should be carried out through the osteotomy between the buccal and lingual cortices. Although wide periosteal stripping is inevitable to allow visualization for ramal osteotomies, the surgeon must exercise care to expose only what is necessary.

Neurologic Complications

With the exception of the nasopalatine and superior alveolar nerves that are inevitably transected with the LeFort I osteotomy, the sensory loss of the infraorbital nerve distribution is temporary; there is nearly complete recovery, because the nerve is well visualized, and the vast majority of the osteotomies are inferior to the foramen. The long-term incidence is approximately 1.5% to 2%, and it may occur as a result of extensive traction from soft tissue retraction or compression at the time of plate fixation.^{109,129,130} The greater palatine nerve is frequently transected with the posterior osteotomy but without apparent significant sensory disturbance to the patient. Sensory deficits of the teeth and palatal and buccal mucosa seem to gradually resolve over a 12- to 18-month period. Other rare neurologic deficits of the second, third, fourth, fifth, sixth, tenth, and twelfth nerves have also been reported.¹³¹⁻¹³⁴ These are likely to occur as a result of unfavorable fractures ascending into the cranial base.

In contrast, the sensory deficit is the major concern with mandibular osteotomies, because the nerve is not visible and the surgeon relies primarily on anatomic data that are not specific to individual patients.^{112,113,135} Of the mandibular procedures, the sagittal split osteotomy of the ramus has the greatest risk of sensory loss.^{116,136} The inferior alveolar nerve is at risk at virtually every point during the sagittal split osteotomy procedure. The nerve is at risk with exposure of the medial ramus, the horizontal osteotomy of the medial ramus, the vertical osteotomy of the buccal cortex and the osteotomy along the external oblique ridge, the splitting of the ramus between the proximal and distal segments, and with fixation. Despite all precautions, a significant

percentage of patients will have sensory loss with or without transection. Transection that is apparent at the time of surgery is reported to range from 1.3% to 18%.^{102,115,135,137} Even without transection, long-term sensory loss in the inferior alveolar nerve is reported to occur in 24% to 85% of cases.^{112,113,135,138-140} Correlation between the surgeon's intraoperative assessment of the nerve injury and postoperative neurosensory deficit is poor; thus reacting to a perceived nerve injury that does not include complete transection is in most circumstances not indicated.^{141,142}

Other neurologic deficits have been reported with the sagittal split procedure.¹⁴³ The lingual nerve may be affected by the osteotomy of the inferior border, especially when an inferior border saw is used. Seventh nerve palsy is uncommon but has been reported to occur in 0.4% to 1% of patients because of the nerve's close proximity to the posterior border of the ramus.^{112,113,144} Unlike the sagittal split osteotomy, the risk of nerve injury with IVO is theoretically lower, although long-term sensory deficits range from 2.3% to 14%.^{116,136,145} Again, this is attributed to the inability to directly view the foramen located medially and placement of the vertical osteotomy well posterior based on lateral landmarks. Despite the significant percentage of neurosensory loss, patients are rarely bothered, and few, if any, mention it unless the surgeon asks. However, there should be a frank discussion with the patient before surgery, and if the patient cannot accept the possibility of sensory loss as a tradeoff for the benefits of occlusion, mandibular surgery should not be offered.

With osseous genioplasty, superficial distal fibers of the mental nerve are transected with the mucosal incision, and the mental nerve is at risk with the proximity of the osteotomy. Ideally, the osteotomy should be placed 4 to 5 cm below the foramen, because the nerve within the canal descends below before exiting the foramen. There may be insufficient bone below the foramen to safely carry out the osteotomy at this level, and the osteotomy is, of necessity, made proximal to the mandibular border, resulting in discontinuity when the symphysis is repositioned. At times, extending the mucosal incision or a second posterior exposure of the inferior border will facilitate visualization and the osteotomy. Despite precautions, the long-term sensory deficit has been reported to be as high as 20%.^{139,146,147} When osseous genioplasty is combined with a BSSO, as is frequently done, the long-term sensory deficit significantly increases from approximately 10% with genioplasty and 30% with a sagittal split osteotomy alone to 70% when a combined procedure is performed.^{126,133}

Infection

Postoperative surgical infection is uncommon after orthognathic surgery; the incidence is less than 1%.^{102,112,148-150} As with most operative procedures, the risk of infection increases with the length and type of the operative procedure. Bacterial contamination is inevitable, and the use of perioperative prophylactic antibiotics is common.^{148,151,152}

Unfavorable Osteotomies

With the LeFort I procedure, uncontrolled fractures extending into the cranial base occur when the osteotomies are incomplete and downfracture is attempted or when the pterygopalatine disjunction is improperly positioned, resulting in uncommon vascular, neurologic, and ophthalmic complications.¹⁵³⁻¹⁵⁵ The downfracture should occur with digital pressure alone, and Rowe disimpaction forceps should be used only for mobilization after the maxilla is separated from its base. Frequently, with digital pressure, sites of resistance can be identified, and these tend to occur at the posterior aspect of the lateral nasal wall and the posterior maxillary wall where the reciprocating saw was not fully positioned at its maximal depth. A thin osteotome can be introduced through the osteotomy of the anterior wall until resistance is felt. The osteotome should

be directed inferiorly from the orbital floor and posterior wall perforated with downward digital pressure until the maxilla downfractures. Whereas in developmental dentofacial deformities the maxilla readily downfractures because the posterior maxillary wall is thin and readily fractures favorably, in cleft and craniofacial cases the posterior wall is excessively thick, and an osteotome may be needed to fully complete the osteotomy. Although the traditional LeFort I osteotomy readily downfractures, the downfracture of the higher level LeFort I osteotomies that include the body of the zygoma does not readily occur and requires the surgeon to direct the osteotomy of the posterior lateral maxillary wall from the pterygoid plate superiorly with an osteotome. Visualization is difficult and requires an understanding of the 3D skeletal anatomy.

Unfavorable fractures from sagittal split osteotomies of the ramus occur with an incidence of 3% to 23%.^{102,113,115,137} These include condylar neck fractures, lingual plate fractures, and buccal plate fractures. The ideal split may be technically difficult to achieve, depending on the anatomic variations and ramal width and with it the ability to place and direct the osteotomes.¹¹³ Condylar neck fractures occur when the horizontal osteotomy is misdirected posterosuperiorly instead of horizontal to the occlusal plane. Buccal plate fractures typically occur when the vertical osteotomy at the inferior border is incomplete and a sagittal split with the osteotome is attempted. Lingual plate fracture may occur, typically originating near the ascending ramus and when the mandibular third molars have not been previously extracted. Unerupted third molars should be extracted 6 to 12 months before surgery to avoid uncontrolled fractures and allow ease of internal fixation.¹⁵⁶ Depending on the fracture pattern and whether there is advancement or setback, the surgery may proceed but will require principles of fracture management and prolonged postoperative maxillary-mandibular fixation.¹⁰² Alternatively, the procedure may need to be abandoned and the fractures allowed to heal before returning to the operating room.

Postoperative Malocclusion

Surgical malocclusion is uncommon but can occur and becomes readily evident in the early postoperative period.^{102,156-158} An early tendency to relapse can frequently be managed with class III or class II dental elastics and/or orthopedic appliances if minimal (edge-to-edge incisal relation). However, more significant recurrences of the initial preoperative occlusion in most circumstances necessitate a return to the operating room. In most cases, early failure results from inadequate mobilization of the repositioned jaws, bony interferences, and instability that is not detected during repositioning, condyles not passively seated within the glenoid fossa at the time of fixation, and issues related to internal plate-screw fixation. In addition, occlusal relapse can occur long after the initial surgery and is a result of complex functional dental and muscular forces reestablishing equilibrium and remodeling of the facial skeleton. Long-term follow-up is important, and management must be directed at the cause.

OUTCOMES

Unlike many surgical procedures, the outcome depends not only on the surgical procedure itself but also on a multitude of factors that exist long before the surgery, as well as on the ability to control variables long after the surgical procedure. Whether the desired long-term outcome is achieved in terms of occlusal function and aesthetics of the facial form depends on whether the goals of each of the phases of treatment were achieved. Moreover, the success of each phase depends on the preceding phase of treatment. For example, inadequate incisor decompensation limits the amount of sagittal repositioning possible and compromises the final facial aesthetics.

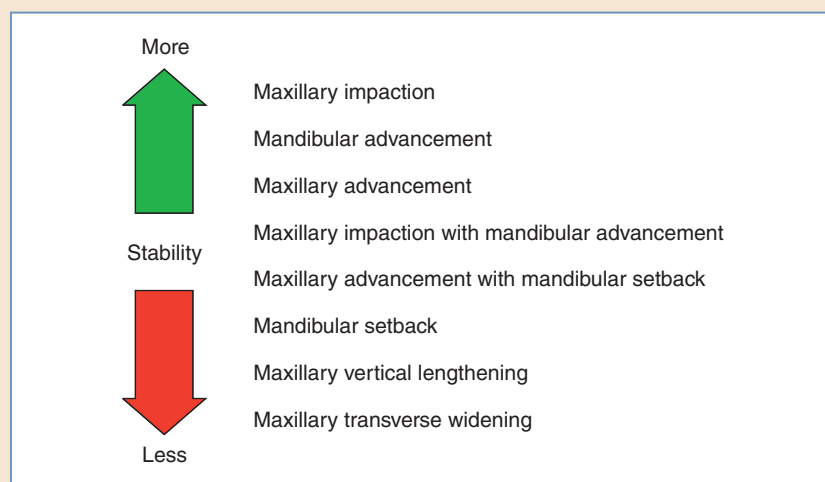


Fig. 27-65 A hierarchy of skeletal stability based on the vector of repositioning the maxilla and mandible.

If mobilization of the maxilla at the time of surgery is inadequate with a less than ideal occlusal relation obtained, the postsurgical orthodontic phase is prolonged and relapse toward the original condition is likely to occur.

With any skeletal movement, the surgeon must always be aware of the potential for relapse, even under an ideal setup and with the use of rigid internal fixation. As a general rule, the most stable moves are superior maxillary impactions, followed by mandibular advancement^{159,160} (see Fig. 27-63). In contrast, maxillary vertical lengthening, followed by mandibular setback, are less predictably stable procedures. The least stable is transverse maxillary widening. The stability of the repositioned maxilla and mandible is the subject of numerous publications¹⁶¹⁻¹⁶⁶ (Fig. 27-65). Many factors come into play, such as the specifics of the internal fixation, use of bone grafts, the period of postoperative occlusal stabilization, and amount of repositioning. Soft tissue forces directed against the vector of the surgical movement are significant. Ultimately, long-term skeletal stability requires neuromuscular adaptation.^{159,167,168} With maxillary impaction, the forces from the mandibular elevator muscles are favorable for maintaining the vector of movement, in contrast to maxillary vertical lengthening. Similarly, compensatory repositioning of the tongue to maintain the airway is more favorable with advancement than with setback.¹⁶⁹ Although it may seem reasonable that morphologic stability and physiologic adaptation should be complete 1 year after surgery, skeletal and dental changes can continue to occur.^{160,170,171}

CONCLUSION

Although orthognathic surgery involves restoring the skeletal anatomy, it is ultimately how the soft tissue drapes on the new facial skeleton that is the patient's concern.¹⁷² The surgeon must be well aware of the soft tissue response to the skeletal movements.¹⁷³ The goal is not necessarily to normalize the cephalometric values; the goal is to have the patient appear "normal" and to function "normally." The ability to predict the skeletal and soft tissue changes with orthognathic surgery is critical to treatment planning. With the current surgical techniques that use plate fixation,

the skeletal components can be repositioned with a relative degree of confidence. However, the soft tissue response is less predictable because of the multitude of uncontrolled variables, which include the surgical technique itself, the vectors and extent of repositioning, thickness of the musculature and subcutaneous adipose tissue, and ability of the soft tissue to adapt or conform to the new dentoskeletal morphology.^{163,174-181} Much of our knowledge about the soft tissue response is based on 2D cephalometric and photographic analysis. However, it is the 3D redraping of the soft tissue envelope to the new dentofacial skeletal morphology that requires quantification.¹⁸²⁻¹⁸⁴ Until sufficient data are available and better algorithms can be generated, orthognathic surgery remains an art based on the level of experience of the orthodontist and surgeon.

KEY POINTS

- Orthognathic or maxillofacial surgery should be thought of as a subcategory of craniofacial surgery.
- Indications for orthognathic surgery include facial dysmorphism with or without functional implications.
- Maxillofacial deformities can be broadly divided into three major categories: skeletal dysplasias, dental dysplasias, and dentofacial skeletal dysplasias.
- Correction of maxillofacial deformities requires careful analysis of the soft tissue with clinical examination and supporting photographs, skeletal evaluation with standardized radiographs, and dental evaluation with study models.
- In orthodontic-surgical management, the primary decision that must be made is whether the deformity is significant enough to require surgical repositioning, or whether orthodontic alignment of the dentition alone can be achieved without significantly compromising the facial aesthetics.
- The elements of the facial skeleton can be repositioned, redefining the face through a variety of well-established osteotomies.
- Various osteotomies are used to correct midfacial deformities; the choice of a particular procedure depends on the specific deformity.
- Orthognathic surgery to correct dentofacial deformities in patients with facial clefts is a more complex undertaking, because such patients present with not only skeletal considerations but also the soft tissue component, which plays a significant role in the management and outcome.
- Although orthognathic surgery involves restoring the skeletal anatomy, it is ultimately how the soft tissue drapes on the new facial skeleton that is the patient's concern. The surgeon must be well aware of the soft tissue response to the skeletal movements.

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28

Alveolar Cleft Surgery

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Power Iliac Crest Bone Graft Harvest; Imaging, Surgical Markings for Power Iliac Crest Bone Graft Harvesting, and Placement of Bone Graft in the Alveolar Cleft.



The study and treatment of the alveolar cleft have remained a less characterized and often underappreciated component of cleft care in the plastic surgery literature. The alveolus is the functional keystone in the management of the complete cleft lip phenotype because of its position at the apex of the primary palate and junction with the secondary palate. The ultimate repair of the persistent alveolar bone defect after initial cleft lip and palate surgery is of great importance in properly creating a unified maxilla and solid foundation for future facial skeletal surgery.

The goals of successful bone reconstruction include:

- Optimal dental eruption and salvage of permanent teeth erupting within the cleft
- Creation of a stable one-piece dental and maxillary arch
- Proper skeletal support for the nose
- A durable substance between oral and nasal lining repairs, decreasing the chance for recurrent oronasal fistula
- Creation of adequate bone stock for future dental restoration with osseointegrated implants or orthodontic canine substitution

This chapter will focus on the comprehensive management of the alveolar cleft defect from infancy (deciduous dentition) through mixed dentition and finally with permanent dentition while highlighting the advances in bone biology that have improved on results during the first decade of the twenty-first century.

German surgeons Lexar, Drachter, and von Eiselbergin¹ first reported the repair of alveolar and maxillary defects with bone grafts in cleft patients in the early 1900s. Although described nearly 100 years ago, reconstruction of the maxillary and alveolar defects in cleft patients was rarely practiced until Boyne and Sands² reported their experience in 1972 with secondary autograft bone grafting of palate and alveolar defects in 12 cleft patients in mixed dentition. Currently, autograft iliac crest bone remains the most commonly used graft material. Yet, significant donor-site morbidity, including meralgia paresthetica, hematoma, intestinal herniation, infection, and postoperative fractures, persists with this approach.^{3,4}

Significant controversy remains regarding the optimal management of the cleft alveolus, including optimal timing of surgery (infancy, mixed dentition, or permanent dentition), the type and location of osseous grafting material, and the optimal design and transposition of vascularized tissue to promote graft material healing and minimize future dental complications, such as gingival recession. The plethora of treatment options and the paucity of comparative outcome data arguably define the cleft alveolus as the least studied and most enigmatic aspect of comprehensive cleft care.⁵⁻¹⁶ This chapter will explore each of these critical controversies and provide reasonable treatment recommendations for the treatment of the cleft alveolus.

EPIDEMIOLOGY AND EMBRYOLOGY

The incidence of the cleft alveolus mirrors that of the common cleft lip and varies greatly based on race and ethnicity. In white populations, the incidence has been reported as high as 1 per 1000 live births and as low as 0.41 per 1000 live births in populations of African descent.¹⁷ Asian populations, including the Indian subcontinent, have rates twice as great (2 per 1000 live births).¹⁷ Interestingly, the incidence of clefting isolated to the secondary palate has been reported at 0.5 per 1000 live births and similar among various ethnic and racial groups.¹⁸⁻²⁰

Most children born with a cleft lip, including the primary palate, are males, whereas isolated clefts of the secondary palate typically occur more commonly in females.²⁰ The incidence of unilateral cleft lip occurs twice as often on the left side compared with the right side. Unilateral cleft lip and primary palate occurs more frequently than bilateral cleft lip at a ratio of 9:1.²⁰ A notable phenomenon is isolated bilateral clefts of the lip only that more commonly present with an associated cleft of the secondary palate in 86% of bilateral cleft cases. In contrast, a unilateral cleft lip is diagnosed with a concurrent cleft of the secondary palate in 68% of cases.²⁰

The pathogenesis of facial cleft has generally been thought to result from a failure of fusion of any of the five developing facial elements that include the frontonasal process, the two lateral maxillary processes, and the two mandibular processes of the embryo. By the fourth week of gestation, neural crest cells from the ectoderm proliferate and migrate to form a substantial portion of the facial mesenchyme. By the end of the fifth week, the maxillary prominences have enlarged and merged with the lateral nasal prominences, establishing continuity between the nose and cheek and have fused with the medial nasal prominences, establishing continuity of the lip. Failure of any one or both of the maxillary prominences to merge with the fused medial nasal prominences results in a unilateral or bilateral cleft of the lip, respectively. Plate formation begins during the fifth week of gestation and is completed by the twelfth week of gestation. Failure of fusion of the median palatine process that is derived from fusion of the median nasal processes results in creation of the primary cleft palate or cleft alveolus. The primary palate, when properly formed, will become the premaxillary segment of the upper jaw. This segment ultimately gives rise to the alveolar structures, including the dentition and portion of the hard palate anterior to the incisive foramen.²⁰

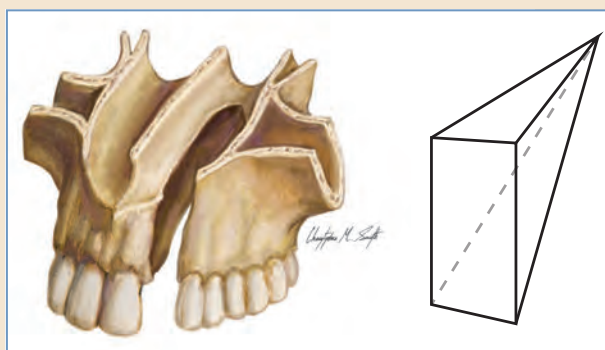


Fig. 28-1 A cleft alveolus demonstrating its shape, resembling a platonic solid or tetrahedron. The apex of the shape corresponds to the incisive foramen. The cleft represents a discontinuity of the maxilla into two pieces in the unilateral cleft lip. As suggested by the figure, routine cleft repair without gingivoperiosteoplasty does not address the bone defect.

NORMAL AND ABNORMAL ANATOMY

The *alveolar cleft* is a complex defect that has a shape resembling a regular convex polyhedron or platonic solid, such as the tetrahedron when fully developed before reconstruction with bone graft, and is a critical component of the complete cleft lip deformity (Fig. 28-1). In an isolated cleft of the secondary palate, the maxillary alveolus is complete and intact. In the *unilateral complete cleft lip*, the maxilla is separated into a major or greater segment, usually on the cleft side, and minor segment opposite. The vomer bone is usually fused to the major segment and exposed on the side of minor segment within the oronasal cleft defect. In *bilateral clefts*, however, the vomer bone and associated mucosa are exposed on both sides and fused to the premaxilla, creating an unfavorable condition in which vomer-directed growth is unrestrained or guided by either lateral maxillary segment (fly away premaxilla). Whether unilateral or bilateral, the alveolar cleft is lined by both nasal and oral mucosa that ultimately becomes continuous with the buccal mucosa on the labial side, continuous with the palatal mucosa on the palatal side, and contains varying amounts of keratinized gingival epithelium at the anterior cemento-enamel junction.

TIMING OF SURGERY

Significant controversy exists regarding the optimal time for treatment of the alveolar cleft. A significant concern with most bone-related surgery is the requirement for subperiosteal exposure that may ultimately devascularize the adjacent bone and secondarily disrupt or inhibit future facial growth. When alveolar bone graft (ABG) cleft surgery is performed early, before any tooth development, it can be done with bone or cartilage graft (primary ABG) or without grafting material that is known as *gingivoperiosteoplasty* (GPP), which is performed at the time of the primary cleft lip repair. Whether repaired early or late, the cleft alveolus–maxillary bone defect is treated by the multidisciplinary cleft-craniofacial team; the orthodontist first aligns the maxillary segments over several months, followed by surgical alveolar bone grafting, where the bone graft is placed in the alveolar-maxillary gap, covered by vascularized soft tissue, to unite the maxillary segments into one stable alveolar-maxillary complex. When the bone graft survives in this new

Table 28-1 Treatment Options From Infancy to Adolescence

Infancy: Primary Dentition	Childhood: Mixed Dentition	Adolescence: Permanent Dentition
Dentoalveolar molding, primary grafting, gingivoperiosteoplasty	Orthodontics, secondary grafting	Orthodontics, orthognathic surgery, late repair

location, it can facilitate dental eruption and future orthodontic movement of teeth into the new bone to treat potential cleft dental gaps resulting from missing or malformed teeth within the cleft defect.²¹ The bone graft material is most commonly harvested from autologous iliac crest bone graft (ICBG), although anterior tibia rib and cranium are well-reported alternate harvest sites.²²⁻²⁵ ICBG is usually performed during the period of mixed dentition (secondary ABG) between 7 to 12 years of age, whereas primary GPP repair of the cleft lip-alveolus is typically performed between 3 and 6 months of age. Table 28-1 shows current treatment options for the cleft alveolus based on age of the child, and the concurrent dental development highlights primary versus secondary grafting versus late grafting.

Maxillary Growth

Multiple studies have suggested that surgery attenuates maxillary growth. Lateral cephalometric analyses in the 1950s through the 1970s showed that nonoperated clefts and noncleft controls had no differences in sagittal facial growth other than the region of the cleft.²⁶⁻²⁹ These studies demonstrated that children with clefts have the potential for normal facial growth, and that the surgery itself likely affects growth to some degree. Motohashi et al³⁰ demonstrated that unrepaired adult clefts have wider faces (other than their maxilla) than noncleft control subjects, but repaired adult clefts have narrower maxillas than noncleft control subjects. This finding occurred in patients who only were treated with primary lip and palate repairs without specific treatment of the cleft alveolus. This documented concern of negatively impacting maxillary growth with bone grafting is a common reason for the generally accepted delayed treatment of the cleft alveolus until the mixed dentition period, which typically occurs between 7 and 12 years of age.³¹⁻³⁶ When reconstruction is performed during mixed dentition, it is commonly reported as *secondary ABG*.

When alveolar bone grafting is performed before or during deciduous dentition or before the age of 6 years, this is commonly referred to as *primary ABG*. Many authors have reported decreases in maxillary growth with primary ABG.³⁷⁻⁴⁰ However, in a six European center comparison, Molsted et al⁴¹ showed that primary ABG may have the advantage of more symmetrical dentoalveolar development when viewed from the frontal plane. These findings are intuitive, because early repair of the alveolar cleft may cause less tipping of the teeth and the actual maxillary segments on either side of the alveolar cleft. In the same study, the primary ABG group demonstrated improved symmetry but also decreased maxillary growth in the sagittal and transverse planes.⁴¹ Trotman et al⁴² similarly showed improved dental inclination with primary alveolar bone grafting. Both teams used presurgical orthopedics (appliances to align the infant alveolus). There is concern that presurgical orthopedics may negatively affect maxillary growth.

Enemark et al⁴³ followed 224 cleft patients for 4 years. They were separated into three groups: (1) those who received an ABG before canine eruption (mean age 10 years), (2) those who received an ABG after canine eruption (mean age 13.1 years), and (3) those whose ABG was delayed (after 16 years of age). All patients were compared with a control group of patients with nongrafted

unilateral clefts. All their patients had transverse expansion of the maxilla orthodontically before bone grafting. Enemark's team demonstrated that sagittal maxillary growth was not affected by secondary bone grafting when compared with a nongrafted cleft control subject, but that *anterior facial height* (nasion to anterior nasal spine, or N-ANS) showed less growth in patients with a grafted cleft. Maxillary length was shorter in the after canine eruption group compared with the precanine eruption group; both were shorter than that in the control group. Overall, the youngest group (precanine eruption) had the best levels of alveolar bone at the gingival margin.

Collectively, studies related to the maxillary arch suggest that earlier ABG is better for the height of the alveolar bone and symmetry of the frontal plane, but probably negatively affects some aspect of maxillary growth (for example, height and anteroposterior length).³⁶⁻⁴² The studies cited had mixed results with respect to maxillary width (transverse growth), possibly because of variance in the use of maxillary expansion. Maxillary expansion before ABG may have compensated for any detrimental effects of the bone grafting procedure itself on maxillary transverse growth. Many teams prefer to avoid early bone graft and prefer grafting in mixed dentition before the eruption of the canine teeth, because this group had the best outcomes in the study by Enemark et al.⁴³ Despite this, approximately 25% of children with unilateral cleft lip and palate will require LeFort I maxillary advancement surgery at skeletal maturity to gain a normal bite and appearance.⁴⁴

Dentition and Orthodontia

A detailed review of the literature demonstrates that successfully spontaneous eruption of the canine tooth through ABG material varies from 27% to 97% and averages approximately 75%.³⁶ For teeth on either side of the cleft, both primary ABG and secondary ABG showed improved dental inclination.^{41,42} Less known is whether dentition can be damaged by ABG. Early surgery in the cleft area seems to disturb the development of the permanent lateral incisor.⁴⁵ However, Enemark et al.⁴³ found greater risk of loss of teeth from external root resorption only in their two older groups, possibly from an ABG damaging the periodontal ligament or cementum of the root surfaces. The fate of the cleft tooth in the setting of an ABG is not always clear from these studies; Enemark et al.⁴³ rated the functional and aesthetic properties of the reconstructed alveolus to be better in their youngest group (average 10 years) compared with their older groups (average 13.1 years and older than 16 years) for both unilateral and bilateral clefts.

Gingivoperiosteoplasty

GPP is based on the principle of guided tissue regeneration—that subperiosteal exposure of the alveolar edges will allow growth of bone without the need for bone grafting. This is typically performed during one of the infant cleft surgeries, most commonly with the primary cleft lip repair.

In the original Skoog technique, presurgical apposition was not used and was reported to minimally affect facial growth.^{46,47} Other researchers have contested these findings on secondary analysis.⁴⁸ The use of an active presurgical Latham appliance, followed by GPP, was associated with poor maxillary growth.⁴⁹ Passive molding followed by GPP has been reported to not significantly affect facial growth, although only 80% had bone formation, and of those, 40% required secondary bone grafting.⁵⁰ Indications for GPP are well-approximated alveolar clefts, with a limited gap between segments of 1 to 2 mm, and therefore require presurgical infant orthopedics (nasalveolar molding, taping, and lip adhesion), to facilitate alveolar segment alignment before GPP.⁵⁰ Given the distinct timing of primary and secondary grafting, the surgical techniques described in the next section will be discussed separately for either GPP or ABG.

PREOPERATIVE ASSESSMENT

Patient History and Physical Examination

For both GPP and ABG, it is critical that the alveolar segments are well approximated before surgery. In an infant, this may be done with nasoalveolar molding, taping, lip adhesion, or other presurgical orthopedics to approximate the alveolar segments. GPP can be done at the time of cleft lip repair or cleft palate repair, although it is typically done during cleft lip repair.

In ABG, palatal expansion is usually performed before the ABG to align the segments into a smooth, curved arch form. There is controversy regarding timing of palatal expansion before or after ABG or both. Teeth within the cleft alveolar margin may inhibit optimal watertight closure of the vascularized flap and may require extraction 2 months before ABG to allow healing and facilitate subsequent vascularized flap closure of the cleft defect.

An unusually wide alveolar cleft may be treated by first collapsing the defect to create a smaller cleft defect to facilitate closure, followed by expansion 1 to 2 months after repair. Another option is staged soft tissue closure with vascularized flaps, followed by bone grafting 3 months later.

Evaluation

Evaluation begins based on the treatment philosophy of the cleft-craniofacial team. When will the procedure to correct the alveolar defect take place?

- In the neonatal period (primary repair), reconstructed at the time of the cleft lip repair with the use of GPP
- In the mixed dentition period (secondary repair), before the eruption of the canine tooth
- In the permanent dentition period (tertiary repair), after the eruption (or impaction) of the canine tooth

Patients in the last category have often had prior failed treatment either during primary or secondary repair. Team evaluation by the surgeon, pediatric dentist, and the craniofacial orthodontist is necessary to coordinate optimal treatment. The critical factors in developing the treatment plan before alveolar repair are based on the timing of the repair (Box 28-1).

We prefer secondary alveolar bone reconstruction (ABR) during mixed dentition. The evaluation process typically begins by 8 to 9 years of age to minimize patient anxiety typically present when orthodontic treatments are started before the age of 8 years. In addition, eruption of

Box 28-1 Primary, Secondary, and Tertiary Repair Plans

- Primary repair: Alignment of the maxillary-alveolar segments and narrowing the intersegment gap to under 1 to 2 mm to facilitate tension-free repair
- Secondary repair: Alignment of the maxillary-alveolar segments and narrowing of the intersegment gap to under 8 mm and early decisions regarding extraction of unusable teeth or canine substitution into the lateral position in patients missing a lateral incisor
- Tertiary repair: Alignment of the maxillary-alveolar segments and narrowing of the intersegment gap to under 8 mm and definitive decisions regarding extraction of unusable or carious teeth and canine substitution

the permanent central incisors and permanent first molar (6-year molar, because it arises around 6 to 7 years of age) is necessary to use tooth-borne palatal expansion. We follow cleft tooth (lateral incisor or canine) development, but begin expansion when the child can tolerate expansion rather than wait for traditional development of two thirds of the canine root. Our experience suggests that waiting to initiate orthodontic treatment leads to delays in alveolar cleft reconstruction because of the significant time requirements needed for controlled arch expansion. In rare instances bone-borne palatal expansion devices may be necessary because of the inability to maintain expansion with tooth-borne systems or to bond a device to the permanent teeth as a result of delayed or inadequate molar eruption. Finally, in patients with large bilateral alveolar cleft defects, a staged approach reconstructing only one side at a time and separating the second stage by 3 to 4 months has proved a useful treatment strategy in the high-risk case.

Imaging

Imaging with traditional dental radiographs, including the pantomogram and apical and occlusion techniques, has been used extensively for more than 40 years. Imaging scoring systems, including the Enemark and Chelsea scoring systems,⁴³ have used changes based on apical imaging views before and after intervention (Fig. 28-2). Within the past decade, cone-beam computed tomography (CBCT) has emerged as the imaging modality of choice because of the superior imaging quality when compared with apical dental radiographs. In addition, CBCT data can be reformatted with software algorithms to provide a standard cephalogram and pantomogram and therefore theoretically reduce the total radiation dose to the patient without compromising imaging quality or completeness of the dentofacial imaging record. CBCT also provides data to help determine the root development of the cleft tooth (lateral incisor or canine) and measure the dimensions of the alveolar cleft, including volume loss and relative amount of bone graft necessary for complete reconstruction.

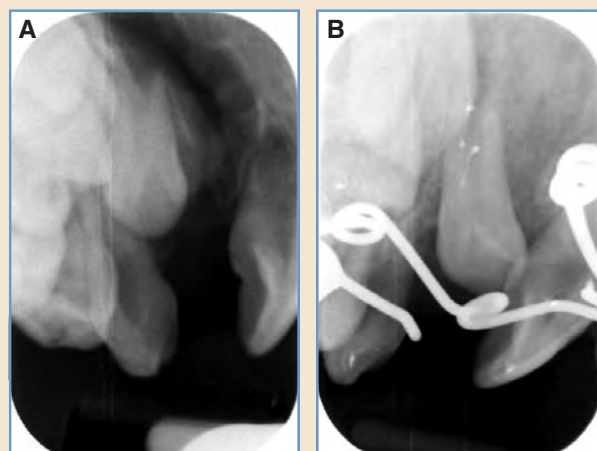


Fig. 28-2 **A**, An apical dental radiograph showing a typical unilateral alveolar cleft defect before treatment with adjacent deciduous and permanent teeth on either side of the cleft bone defect. **B**, An apical dental radiograph showing the same unilateral alveolar cleft region after treatment with our preferred technique of minimal access ICBG and allograft bone graft. Note the complete obliteration of the cleft bone defect and replacement with mature bone that has supported eruption of the adjacent teeth. This is a typical Enemark 1 result.

OPERATIVE TECHNIQUE

Principles of Alveolar Bone Grafting

There are multiple techniques for bone graft reconstruction (Box 28-2) and vascular coverage (Box 28-3).

Complete reconstruction of the alveolar cleft defect requires proper closure of the roof (nasal side), floor (palatal side), and anterior (labial side) gingiva and mucosa of the upper lip. It also requires elevation of a contiguous gingivoperiosteal flap to the first molar on the cleft side (or both in bilateral cases) and to the contralateral incisors on the noncleft side (or not crossing the prolabium to avoid devascularizing the premaxilla in bilateral cases). Alternatively, gingival and buccal flaps can be separately elevated, as reported by Precious.⁵¹

The mucosa overlying the alveolar cleft is divided to create nasal-palatal flaps. We recommend a marginal incision within the arch of the alveolar cleft, whereas others recommend that the incision be placed more labially to allow larger turnover flaps to line the limited nasal floor. We avoid using this technique, because it places nonrespiratory mucosa in the nasal cavity. Regardless, the exposure must be in the subperiosteal plane to fully expose the bone defect and remove any intervening soft tissue that may limit ultimate alveolar fusion. Depending on the size of the alveolar cleft, this exposure may just graft the leading edge of the alveolus, or more typically, needs to reopen and cross the scar tissue of the original oronasal fistula repair (that is, the original primary cleft lip repair) to ensure full takedown and closure of the residual oronasal fistula. This theoretically would only go posteriorly as far as the incisive foramen, because this is the limit of the alveolar cleft. However, in certain patients with unrepaired clefts or large preexisting oronasal fistulas, this dissection may have to proceed further posteriorly.

The nasal flaps need to be reapproximated tightly to avoid persistent oronasal fistula or communication of the nasal cavity with the underlying alveolar graft material. This is typically done with an absorbable suture but may be augmented by other materials, such as allograft dermis (for example, AlloDerm) or tissue sealant (for example, Tisseel). Methylene blue through the cleft nostril may be used to test for the presence of functional leakage through an oronasal fistula.

Box 28-2 Techniques for Bone Graft Reconstruction

- No bone graft: Avoid bone grafting the osteogenic reconstruction with GPP
- Autograft bone graft: Various donor-site locations, although the most common is the iliac crest
- Allograft bone graft: Corticocancellous chips, demineralized bone matrix, or a combination of both
- Biologic agents: Bone morphogenetic protein, platelet rich plasma

Box 28-3 Techniques for Vascular Coverage

- Sliding gingivoperiosteal flaps with or without lateral relaxing incisions
- Transposition local mucosal flaps
- Regional flaps (tongue or facial artery musculomucosal flaps)
- Palatal flaps
- Allograft skin substitutes



Fig. 28-3 Our preferred incision for elevation of gingivoperiosteal upper lip sulcus flaps. Note that medial transposition will ensure that gingival tissue will be placed into its correct position after closure. Placement of buccal tissue in the gingival position may close a fistula but will increase the risk of future tissue recession and ultimate exposure of the cemento-enamel junction.

Depending on the size of the flaps, palatal flaps may need to be elevated to ensure complete floor support of the bone graft. For the largest clefts or those with extensive oronasal fistula, staged repair of the palate floor may be necessary before definitive ABG.

After adequate exposure, full development of the alveolar cleft defect, and closure of the oral and nasal surfaces, the defect is filled with grafting material. An ICBG is typically harvested through an incision lateral and inferior to the anterior superior iliac spine to avoid injury to the lateral femoral cutaneous nerve. Variations include limiting the incision to a stab incision with a trephine (Acumed) to harvest columns of bone.^{7,52} Other authors have described the use of cadaveric bone allograft and bone morphogenetic protein 2 (BMP-2).^{7,53-58} Regardless of the technique, bone graft or bone substitute must be packed against the exposed alveolar bone edges to allow protected regeneration of bone for osteoconduction and likely osteoinduction in fresh bone graft, cadaveric bone allograft (for example, DBX Mix, Synthes), and BMP-2. Some practitioners will decorticate the alveolar edges until bleeding before graft or substitute placement.

Finally, labial soft tissue coverage is typically done with sliding upper lip sulcus flaps, allowing any lateral relaxing incisions to heal by secondary intention. This lateral site should avoid Stensen's duct. Some authors recommend against this technique, because scarring at the lateral donor site can cause periodontal scarring that can lead to problems with future prosthetic bridges or periodontal health, which may necessitate scar release or vestibuloplasty. This problem can usually be avoided if the gingiva is maintained over all the teeth between the cleft and donor site. The advantage of a sliding flap is good restoration of specialized keratinized gingival mucosa, permitting normal dental eruption (Fig. 28-3).

Less commonly, the sliding upper lip sulcus flap may be inadequate for closure, and other variations have been proposed, such as transposition flaps, tongue flaps, or facial artery myomucosal flaps. These flaps have the advantage of bringing greater amounts of vascularized soft tissue to aid in closure of the wide cleft alveolus defect, but the disadvantage is that the mucosa and not the gingiva is transposed into the region of tooth eruption. This lack of replacing gingival tissue in the cleft alveolus may need further treatment with additional gingival grafts or flaps to address recession.

Less Common Variations

Some practitioners recommend that for large clefts, osteotomies should be performed to narrow the bony cleft before ABG. Others will fixate the repair with a corticocancellous bone graft from the iliac crest secured with titanium or resorbable hardware—this would usually be recommended only in the skeletally mature patient or at least after the majority of maxillary growth (older than 12 years of age).

Gingivoperiosteoplasty

GPP requires alveolar bone apposition no more than 1 to 2 mm apart, not including the soft tissue. Mucosal hypertrophy can simulate well-appositioned bony edges, and a careful examination is needed to rule out this false apposition.

The goal of GPP is subperiosteal elevation of flaps to create the ceiling (nasal side), floor (palatal side), and anterior (labial side) aspect of the alveolar space. Ceiling and floor flaps are elevated from the vomer and maxillary segment but leave triangular flaps anteriorly to swing labially. These ceiling flaps, floor flaps, and labial flaps can be approximated with absorbable sutures. Posteriorly, dissection should reach the incisive foramen, which is where the ceiling, floor, and sidewalls merge posteriorly.

Our Preferred Techniques

Donor Bone Graft Harvest—Minimal Access Technique

To minimize autograft bone harvest–related morbidity, minimal access bone harvesting techniques have been developed but can be limited by the volume of graft harvested. At the time of surgery, an ICBG is harvested from the iliac crest with a minimal access incision to facilitate a bone graft harvest of 5 ml of autograft using a power-driven trephine (see accompanying video). The procedure begins with a 1.5 to 2.5 cm incision placed inferior and parallel to the anterior iliac crest. After opening and dividing the subcutaneous tissue, the skin, including Scarpa's fascia, is retracted cephalad over the lateral aspect of the iliac crest. Electrocautery is used to incise the cartilage cap and retracted with an elevator. Next, a starting bone tamp is used to create a concave surface to facilitate harvesting of bone. The same entrance site is used with changing angles of approach to harvest multiple cores of graft. Up to 10 ml of graft material can be reliably harvested without creating a secondary contour abnormality of the donor iliac crest.⁵³

Primary Bone Graft Technique: Autograft and Allograft Bone Reconstruction

After autograft harvest, the donor site is checked for hemostasis; a 0.25% bupivacaine impregnated strip of gelatin sponge such as Gelfoam is placed within the donor-site bone defect for pain control, and the wound is closed in layers. Next, the procedure is transitioned to the oral cavity, where medial and lateral gingivoperiosteal flaps are elevated to expose the anterior aspects of the maxillary and alveolar cleft defect. To completely expose the posterior, medial, and lateral walls of the cleft defect, the cleft alveolar fistula is incised from the incisive foramen to the anterior maxilla (Fig. 28-4, A). The fully defined maxillary defect is shaped as a tetrahedron, extending posteriorly from the incisive foramen to the alveolar ridge anteriorly. The roof of the defect is the nasal closure, and the floor is the palatal closure. The base of the tetrahedron is the anterior labial closure area. The alveolar fistula closure is completed by repairing the nasal and oral mucosal layers separately, creating a large pocket in which to place the bone graft material. Demineralized bone matrix mix, consisting of freeze-dried matrix and cortical chips (DBX Mix), is mixed with the patient's autologous ICBG, bringing the total volume of graft to 10 to 15 ml

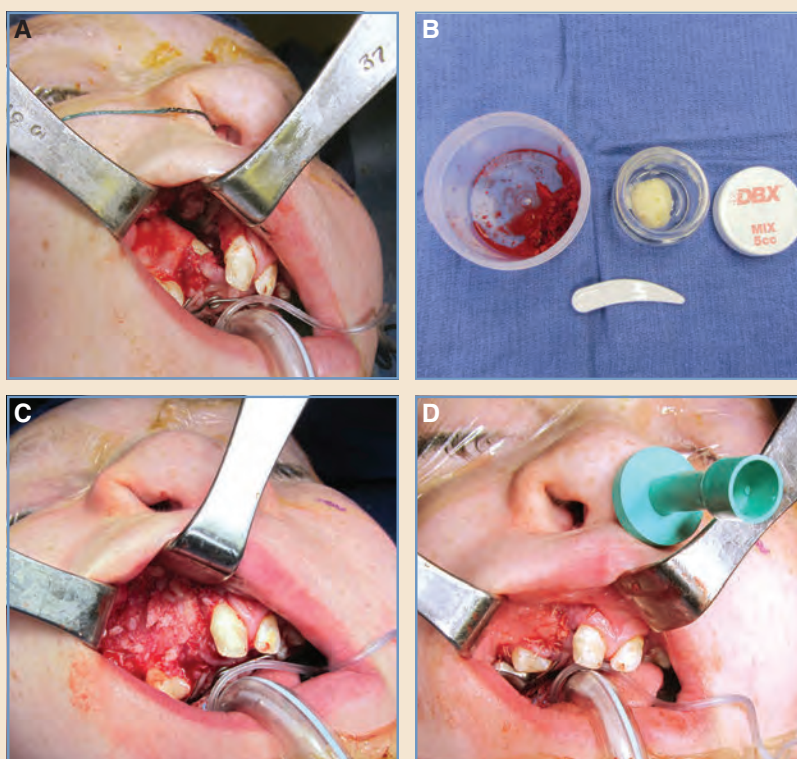


Fig. 28-4 **A**, The fully defined maxillary defect is shaped as a tetrahedron, extending posteriorly from the incisive foramen to the alveolar ridge anteriorly. The roof of the defect is the nasal closure, and the floor is the palatal closure. The base of the tetrahedron is the anterior labial closure area. **B**, Demineralized bone matrix mix, consisting of freeze-dried matrix and cortical chips (DBX Mix), which will be mixed with the patient's autologous ICBG, bringing the total volume of graft material to 10 to 15 ml per side. **C**, The allograft/autograft mix is placed within the cleft defect, including the entire pyramid-shaped defect and piriform rim, after ensuring hemostasis and watertight mucosal closure. **D**, The gingivoperiosteal flaps are reapproximated with suture over the grafted site with incision of the periosteum vertically at the level of the first permanent molar to increase mobility of the lateral flap.

per side (Fig. 28-4, *B*). After hemostasis and watertight mucosal closure are secured, the allograft/autograft mix is packed within the cleft defect, taking care to ensure adequate grafting of the entire pyramid-shaped defect, including the piriform rim (Fig. 28-4, *C*). The gingivoperiosteal flaps are reapproximated with suture over the grafted site, incising the periosteum vertically at the level of the first permanent molar to increase mobility of the lateral flap (Fig. 28-4, *D*). When appropriate, Coe-Pak (GC America) is placed over the incision lines as a surgical dressing.

Revision Bone Graft Technique: Allograft-Only Bone Reconstruction

The revision allograft-only procedure begins and ends in the oral cavity, where medial and lateral gingivoperiosteal flaps are elevated to expose the anterior aspects of the maxillary and alveolar cleft defect (see accompanying video). To completely expose the posterior, medial, and lateral walls of the cleft defect, the residual cleft alveolar fistula requires an incision from the incisive

foramen to the anterior maxilla. The fully defined maxillary defect is shaped as a tetrahedron, extending posteriorly from the incisive foramen to the alveolar ridge anteriorly. The roof of the defect is the nasal closure, and the floor is the palatal closure. The base of the tetrahedron is the anterior labial closure area.

The best way to close the alveolar fistula is to repair the nasal and oral mucosal layers separately, thereby creating a large pocket in which to place the allograft bone graft material. After hemostasis and watertight mucosal closure are secured, demineralized bone matrix mix, consisting of freeze-dried matrix and cortical chips (DBX Mix), is packed in the residual alveolar cleft defect with a total graft volume of 2 to 7.5 ml per side as needed. The bone graft material is packed within the cleft defect, filling the entire tetrahedral-shaped defect up to the border of the piriform rim.

The gingivoperiosteal flaps are reapproximated with suture over the grafted site; care is taken to incise the periosteum laterally and therefore create a lateral relaxing incision at the level of the permanent first molar to increase mobility of the lateral flap and diminish tension at the suture line. When appropriate, Coe-Pak is placed over the incision lines as a surgical dressing.

POSTOPERATIVE CARE

Three main issues arise for postoperative care: (1) maintaining bone position (palatal expansion) while the bone is healing, (2) avoiding injury to the soft tissue flaps, and (3) preventing infection of the grafted material. Maintaining bone position can be achieved with orthodontic support, such as an orthodontic wire or a postoperative splint or expander that does not put pressure on the flaps. Injury to the soft tissue can be avoided by restricting the diet to soft foods for 4 to 6 weeks. The use and efficacy of postoperative oral antibiotics or chlorhexidine mouthwash are currently understudied and stochastic at best. We use perioperative antibiotics, typically a cephalosporin, for up to 1 week because of the large graft volumes placed transorally.

Orthodontic movement and further palatal expansion are usually recommended after 4 to 6 months, which is the typical time for initial bony healing. However, some practitioners recommend earlier movement to stimulate the bone with tension on the bone. Regular clinical follow-up and dental imaging are needed to monitor outcomes, including bone graft volume, dental parameters such as root recession, and other complications.

We recommend that patients be discharged when they can tolerate a full liquid diet, usually within 24 hours. The liquid diet is continued for 2 weeks, followed by a soft diet for 4 additional weeks, and finally a normal diet is allowed at 6 weeks after surgery. A form of alveolar segment stabilization after grafting is ideal with either a tooth-borne splint or maintenance of the existing transpalatal arch. The transpalatal arch can remain in place during surgery but can be removed to facilitate closure. It is replaced postoperatively to prevent premature collapse of the alveolar-maxillary segments.

TREATMENT OUTCOMES AND COMPLICATIONS

Minor complications include suboptimal wound healing, such as wound dehiscence, exposure of bone, and sequestration of nonvascularized bone fragments. These complications can be treated without reoperation, usually by diet restriction and improved oral hygiene, including mouthwash and oral antibiotics. Occasionally, debridement of nonincorporating bone graft material is necessary to support wound healing. In general, wound care and coverage with antibiotic ointment or periodontal dressing (for example, Coe-Pak) will result in healing but often with diminished bone graft volume. Rarely, patients may have infections that require operative debridement.

Outcomes on Autograft/Allograft Bone Grafting

More commonly, outcome variables such as bone graft take and longer-term maintenance can be problematic. Multiple studies have examined bone height, such as the Enemark classification and canine tooth eruption rates, with optimal outcomes in the autograft groups.^{43,59} We have reported our experience with autograft only (group 1), autograft with supplemental allograft (group 2), and allograft only in revision cases (group 3).⁷ We achieved favorable results with our preferred technique that uses minimal access bone grafting with supplemental allograft in first-time bone graft reconstruction cases and an allograft-only reconstruction in revision bone graft cases.⁷ We also demonstrated superior results when compared with traditional open ICBG bone grafting of the cleft alveolus. In group 1, ICBG alone, 22 patients (12 males and 10 females; average age 10 years) were treated between October 2004 and December 2006; 17 clefts were unilateral and 5 were bilateral ($n = 27$). In group 2, ICBG plus allograft, 14 patients (8 males and 6 females; average age 9 years) were treated between August 2009 and August 2011; 6 clefts were unilateral and 8 were bilateral ($n = 22$). In group 3, allograft alone, 11 patients (6 males and 5 females; average age 13 years) were treated between February 2011 and January 2012; 7 clefts were unilateral and 4 were bilateral ($n = 11$). Average follow-up was 47.5, 12, and 3.5 months, respectively. Average operative time per alveolus was 78 minutes for group 3, compared with 147 and 111 minutes for groups 1 and 2, respectively ($p < 0.0005$).⁵³

When group 3 was compared with groups 1 and 2, the average radiographic follow-up was 47.5, 20.4, and 13.3 months for groups 1 through 3, respectively. Average operative time per alveolus was 78 minutes for group 3, which was shorter than the 147 and 111 minutes for groups 1 and 2, respectively ($p < 0.0005$). Average engraftment reported by Enemark scores was 1.0 in group 3, which was better than the 2.0 in group 1 ($p < 0.005$) and similar to the 1.1 for group 2 ($p > 0.05$). Revision ABG with allograft alone improved Enemark scores from 3.7 preoperatively to 1.2 postoperatively ($p < 0.0001$). Hospital stay was 0.9 day for group 3 compared with the 2.4 and 2.7 days for groups 1 and 2, respectively ($p < 0.0001$). Bone graft extrusion occurred in six patients (27.3%) in group 1, no complications occurred in group 2, and a single necrotic central incisor was lost at the time of revision bone grafting in group 3 (9.1%). No wound infections, bleeding requiring transfusion/reoperation, or deaths occurred.⁵³ Table 28-2 provides a summary of our autograft outcomes.

Potential cost increases that were incurred with the use of DBX Mix can be offset by the decreased operative time with our allograft-only technique. The average cost of 5 ml of DBX Mix is approximately \$1000. Operating room costs vary widely and have been reported in the literature to vary from \$21 to \$133 per minute and average \$62 per minute. On average the use of our technique saved 69 minutes ($69 \text{ minutes} \times \$62/\text{min} = \$4278$) per alveolus compared with

Table 28-2 Summary of Autograft Outcomes

Group	Number of Patients	Preoperative Enemark Score	Postoperative Enemark Score	Radiologic Follow-Up	Operative Time/Alveolus (minutes)	Complication Rate (percentage)	Length of Hospital Stay (days)
1: ICBG only	22	4.0	2.0	47.5	147	27.3	2.4
2: ICBG plus allograft	14	4.0	1.1*	20.4	111	0.0*	2.7
3: Allograft only	11	3.7	1.2*	13.3	78*	9.1*	0.9*

*Statistically significant data are in boldface with an asterisk.

traditional ICABG techniques. This represents a potential cost savings of \$3278 ($\$4278 - \$1000 = \3278) per bone graft.⁵³

Outcomes on Biologics

BMP-2 is a commercially available recombinant human protein that induces bone formation where it is implanted, thereby avoiding the need to obtain a bone graft from a donor site.⁶⁰ BMP-2 is potentially advantageous in children, because the small size of the patient decreases the available volume of donor bone graft, reduces the duration of surgery, and eliminates the risk of donor-site complications, such as pain, scarring, or hematoma. More tantalizing, if BMP-2 is placed during one of the other infant cleft surgeries, this could potentially eliminate any future ABG procedures, saving the child from a major operation. Unknown potential disadvantages include impinging maxillary growth that may require jaw surgery to correct or a different clinical bone quality affecting dental eruption or orthodontic tooth movement. The present literature documenting the use of BMP-2 for ABG is plagued with small numbers of patients without control group treatment. We have identified at least five pediatric cleft studies in the literature using PubMed. In 2009 Chin⁶¹ reported pediatric use of BMP-2 on 37 cleft lip and palate repairs, as well as on 6 atypical clefts. All but one patient had successful repairs based on clinical and radiologic examination anywhere between 6 and 25 months. A 2007 study by Herford et al^{62,63} followed 10 patients 4 months after BMP-2 ABR. The percentage of the cleft volume filled with bone postoperatively averaged 71.7% compared with 78.1% in iliac crest autologous bone graft (ICABG) control subjects. Fallucco and Carstens⁵⁶ reported 16 of 17 patients treated with BMP-2 showed increased alveolar bone density as measured in Hounsfield units (HU). The 6-month average postoperative increase was 637 HU versus -83 HU preoperatively.

In 2010 Alonso et al⁶⁴ reported a controlled study of eight patients (ages 8 to 12 years) treated with ICABG versus BMP-2 ABR. CT scans were done 6 and 12 months after surgery. At 6 months, the BMP-2 group showed a bone-filling volume percentage of 59.6% compared with 75.4% for the autograft group. The volume percentage differences between the BMP-2 and autograft groups became statistically insignificant at 12 months, with 75% and 80% reported, respectively. Bone height measurement remained shorter at both the 6- and 12-month intervals. At 6 months, the BMP-2 group had 8.5 mm height versus 13.5 mm height for the control group. Although the difference narrowed at 12 months, bone height for the BMP-2 group was 10.2 mm, whereas it was 13.9 mm for the control group.⁶⁴

In 2008 Dickinson et al¹⁰ published a report describing the use of BMP-2 ABG in adolescent patients (average age 16 years). Nine patients underwent BMP-2 ABR and 11 patients had traditional ICABG. They were analyzed at 1 year with films and CT scans. The authors used a 4-point system based on radiographic fill of the alveolar defect and a 4-point system based on intraoral alveolar ridge healing and alar base augmentation; periapical films used a 5-point system (0 to 4) to rate the bone mineralization among cleft tooth roots. The BMP-2 ABG group scored better than the ICABG group in each of these assessments.¹⁰

In 2013 Francis et al⁵⁷ described the use of BMP-2 encased by demineralized bone matrix putty scaffold (Progenix; Medtronic) in 36 patients and compared them with 19 patients who received traditional ICABG ABG. They found that patients undergoing BMP-2 ABR had 97% successful implantation compared with 84% for patients with ICABG ABG. In the initial BMP-2 ABR group (they separated revision BMP-2 ABR), there was increased radiographic evidence of bone. Finally, the BMP-2 ABR group had less infection and shorter operative times than traditional procedures.⁵⁷

BMP-2 outcomes will need to be studied longer term to answer the question of whether its induced bone formation will provide similar outcomes as autograft and allograft. In addition, timing could be of even greater concern, given that recent animal studies have suggested BMP-2 may induce premature fusion of growth sutures in the juvenile developing skeleton.⁶⁵ Also, reports in spine surgery have found unpredictable location and degree of bone formation.⁶⁶⁻⁶⁹ Therefore BMP-2 may adversely affect growth even more than traditional non-BMP-2 ABR. The senior author (A.Y.L.) was recently awarded a national grant examining a large series of patients at different ages who received BMP-2 ABR, and the results are still pending.⁷⁰

CONCLUSION

Optimal reconstruction of the cleft alveolus remains understudied and at times a technically challenging procedure. With the current controversies regarding timing, choice of bone reconstruction material, choice of soft tissue reconstruction, and complexity of measuring skeletal, dental, and orthodontic outcomes, careful long-term analysis of outcomes will ultimately aid in choosing the most optimal reconstructive algorithm.

KEY POINTS

- The alveolus is the keystone of the complete cleft defect: it is the apex of the maxilla, the site requiring dental restoration, and the region of remaining oronasal fistula after primary cleft lip repair. Successful bone reconstruction of the alveolar cleft will unite the maxilla for future facial skeletal surgery, support dental growth and orthodontic treatment, and close the remaining oronasal fistula.
- The timing of alveolar reconstruction remains controversial, but most studies point to the period of mixed dentition (ages 7 to 9 years) as a reasonable balance between impairing maxillary growth and allowing dental eruption and development through adequate bone. A cleft-craniofacial interdisciplinary approach with the cleft surgeon, pediatric dentist, and craniofacial orthodontist is essential for the preoperative preparation of the patient for surgery and the postoperative use of the new bone.
- Successful ABR requires an understanding of the three-dimensional nature of the defect (platonic solid structure) to create adequate vascularized flaps to line this tetrahedron. Multiple techniques exist for bone reconstruction, including autologous bone graft (autograft), autograft with allograft, allograft alone, and in the future the possibility of combination with biologics.

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29

Dental Principles in Maxillofacial Surgery

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Pediatric maxillofacial surgeons must understand occlusion and the role of dental specialties in maxillofacial surgery. Many oral problems seen in the pediatric population can be treated nonsurgically or be resolved when dental specialists are involved in a patient's care. This chapter will focus on the role of dental specialists in optimizing maxillofacial rehabilitation in pediatric patients. We use case examples to demonstrate the importance of dental records to establish the correct diagnosis. Accurate identification of the underlying problem is crucial to develop an appropriate treatment plan, and plastic surgeons must be well informed about which specialists are available to help them achieve the best results for each patient.

BASIC DENTAL TERMINOLOGY

Table 29-1 lists some commonly encountered dental terms and their definitions.

Table 29-1 Common Dental Terms

Term	Definition
Apertognathia	An occlusion characterized by a vertical separation of the maxillary and mandibular anterior teeth, frequently described as <i>anterior open bite</i>
Articulator	A mechanical device that represents the temporomandibular joints and jaws, to which the maxillary and mandibular casts may be attached
Buccal	Pertaining to or adjacent to the cheek
Cast	A plaster replica of the teeth and surrounding tissues; also known as a <i>model</i>
Centric occlusion	The relationship of opposing occlusal surfaces that provide the maximum planned contact or intercuspation
Centric relation	The relationship of the mandible to the maxilla when the condyles are in their most anterosuperior, unstrained positions in the glenoid fossa
Cephalometric radiograph	A lateral radiograph of the head made with precise reproducible relationships between x-ray source, subject, and film
Class I occlusion	The mesiobuccal cusp of the first permanent maxillary molar occludes in the buccal groove of the permanent mandibular first molar
Class II malocclusion	The mesiobuccal cusp of the first permanent maxillary molar occludes mesial to the buccal groove of the permanent mandibular first molar
Class III malocclusion	The mesiobuccal cusp of the first permanent maxillary molar occludes distal to the buccal groove of the permanent mandibular first molar
Distal	Away from the median sagittal plane of the face and following the curvature of the arch
Labial	Pertaining to the lip, especially in reference to the surface of a tooth
Lingual	Pertaining to the tongue, especially in reference to the surface of a tooth
Mesial	Situated toward the midline of the dental arch
Palatal	Pertaining to the palate, especially in reference to the surface of a tooth
Proclination	Anterior angulation of the anterior teeth
Prognathic	Forward position of the mandible in relation to the cranial base
Retroclination	Posterior angulation of the anterior teeth
Retrognathic	The condition of a mandible that is posteriorly positioned in relation to the cranial base

DENTAL SPECIALISTS

Orthodontist

Orthodontics is the dental specialty that focuses on establishing alignment of the teeth and jaws consistent with a balanced facial pattern. Orthodontists have an important role in diagnosis and treatment planning to correct dentofacial deformities and other craniomaxillofacial anomalies. The orthodontist's role is to orthodontically align the teeth and orthopedically position the jaws in a manner that facilitates surgical correction and optimizes occlusion and aesthetics.

Prosthodontist

The prosthodontic subspecialty of maxillofacial prosthetics involves treatment beyond that of the teeth and supporting structures to include prosthetic restoration of the head and neck. Maxillofacial prosthetics is the branch of prosthodontics concerned with the restoration and/or re-

placement of the stomatognathic and craniofacial structures with prostheses that may or may not be removed on a regular or elective basis. Prosthetic treatment can be classified as (1) intraoral, (2) extraoral, and (3) combination defects. Artificial materials are used to fabricate both extraoral and intraoral maxillofacial prostheses.

Maxillofacial prosthodontists are crucial to rehabilitate patients with congenital, posttraumatic, and oncologic abnormalities. A maxillofacial prosthodontist should be consulted at the time of diagnosis to correct or minimize deficiencies in mastication, swallowing, speech, control of saliva, and aesthetics. When surgery and a prosthetic restoration are required, the plastic surgeon should refer the patient for prosthetic evaluation preoperatively to determine the type and design of the restoration based on the configuration of the defect. Optimal prosthetic reconstruction frequently depends on the maxillomandibular relationship and availability of adequate bony support. A team approach to treatment planning is the most efficient method to achieve a favorable result for the patient.

Pediatric Dentist

A pedodontist is a pediatric dentist who maintains the health of primary dentition and prepares for proper eruption of permanent dentition. Some pedodontists also have training in the management of early orthodontic problems. If a child sustains an injury to a primary or permanent tooth, a pedodontist is the most appropriate dentist to manage and follow the patient's injury.

Oral and Maxillofacial Surgeon

Oral and maxillofacial surgeons have surgical expertise with the mouth and jaws. These surgeons frequently perform osteointegrated implant placement, orthognathic surgery, temporomandibular joint surgery, and distraction osteogenesis. Collaboration between an oral and maxillofacial surgeon and the plastic surgeon may be beneficial in select cases.

Endodontist

Endodontists treat injuries to nerves in teeth. The pulp is the neurovascular tissue in the crown of the tooth (the visible portion). The root of the tooth also has neurovascular tissue that goes from the pulp to the apex of the tooth (the end of the root). Injuries may occur as a result of trauma, osteotomies, tumors, or infections. If a tooth has been injured, it may become sensitive to hot, cold, and percussion. An endodontist can determine whether this is a temporary or permanent condition, and if permanent nerve damage exists, a root canal may be performed. This requires removing the damaged nerve tissue from the tooth and placing gutta-percha. After a root canal is performed, a prosthetic crown is made to cover the tooth and provide strength.

Periodontist

Periodontists specialize in treating the bone that supports the roots of the tooth. Inflammation, trauma, postablative defects, alveolar bone grafts, and other surgical sequelae may alter or compromise the bone that surrounds these roots (periodontal bone). Ideally, attached gingiva is on the alveolar ridges and supports each tooth. In trauma cases it is often necessary to approximate loose mucosa over the alveolar ridge to close the wound. The periodontist can perform gingival grafts to restore the normal presence of attached gingiva in these regions. This reconstruction of normal soft tissue anatomy is very important for long-term maintenance of the dentition. Periodontists typically treat adult patients, but in patients in whom atypical periodontal disease exists, it is appropriate to seek a periodontist for a child.

CONCEPTS OF OCCLUSION

Much orthodontic treatment is based on occlusal concepts first described by Edward Angle¹ in the late 1800s. Angle classified occlusion based on the maxillary first molar and its relationship to the mandibular first molar. He described normal occlusion as class I—that is, the mesial buccal cusp of the maxillary first molar occludes in the buccal groove of the mandibular first molar without crowding, spaces, or rotations. Angle's concept of normal occlusion remains accepted today and is the primary occlusal relationship desired in most modern orthodontic cases. It is evident that if the molar relationships are ideal, and the teeth are of proportionate size, the remaining dentition will align in an ideal relationship. Whether class I occlusion should be considered “ideal” is a topic of continuing debate. However, most investigators find this relationship to be the most aesthetically pleasing and “protective” of the dentition, because it minimizes aberrant forces. (Fig. 29-1, *A*)

Angle also classified abnormal occlusal relationships, and these classifications of malocclusion are defined as follows:

Class I: Ideal first molar relationship with malposed, poorly spaced, or rotated teeth (Fig. 29-1, *B*).

Class II: Mandibular first molar positioned distally (posterior) to the maxillary first molar.

The maxillary canine cusp is anterior to the mandibular premolar and canine² (Fig. 29-2).

Class III: Mandibular first molar positioned mesially (anterior) to the maxillary first molar.

The maxillary canine cusp is posterior to the mandibular premolar and canine² (Fig. 29-3).

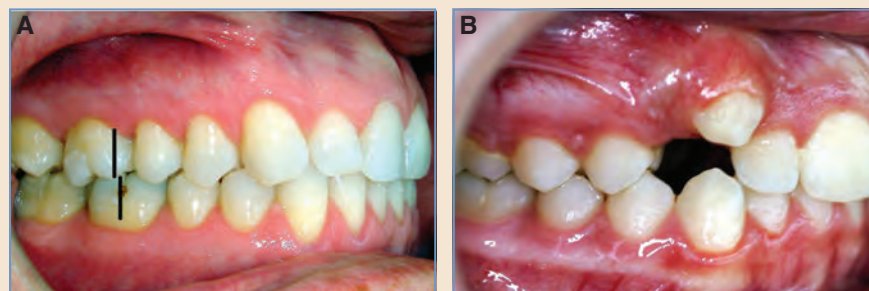


Fig. 29-1 *A*, Class I ideal occlusion. The mesiobuccal cusp of the maxillary first molar occludes in the buccal groove of the mandibular first molar. *B*, Class I malocclusion.



Fig. 29-2 Class II malocclusion. The mandibular first molar is positioned distal (posterior) to the maxillary first molar.



Fig. 29-3 Class III malocclusion. The mandibular first molar is positioned mesial (anterior) to the maxillary first molar.

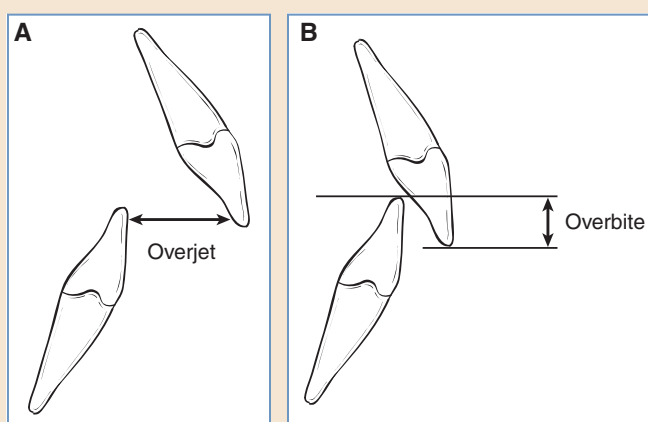


Fig. 29-4 **A**, Overjet. The distance between the labial surface of the lower incisor and the labial surface of the upper incisor. **B**, Overbite. The amount of vertical overlap of the incisors.

Although Angle's classification is useful to describe dental relationships, additional clinical descriptions can help to define occlusion. *Overjet* measures the distance between the labial surface of the lower incisor and lingual surface of the upper incisor (Fig. 29-4, *A*). Ideally, the mandibular incisors contact the lingual surfaces of the maxillary incisors at or above the dental cingulum. This measurement often corresponds to Angle's classification. For example, an ideal class I occlusion has 2 to 3 mm of overjet, which is the thickness of the incisal edges of the anterior teeth. A class II malocclusion will likely display excessive overjet, with the maxillary incisors anterior to the mandibular incisors, and a class III malocclusion will likely display a negative overjet, with the mandibular incisors anterior to the maxillary incisors (see Figs. 29-1 through 29-3).

Vertical dental relationships are described in terms of *overbite*. Overbite is the amount of vertical overlap of the incisors (Fig. 29-4, *B*). As previously stated, the incisal edges of the mandibular incisors should contact the lingual surfaces of the maxillary incisors at or above the cingulum, which equates to 2 to 3 mm of overbite. If there is no overlap of the incisors, this is referred to as an *anterior open bite* (apertognathia) and is measured in millimeters of separation² (Fig. 29-5).



Fig. 29-5 Anterior open bite. There is no vertical overlap of the incisors.



Fig. 29-6 Deep bite. There is excessive vertical overlap of the incisors.

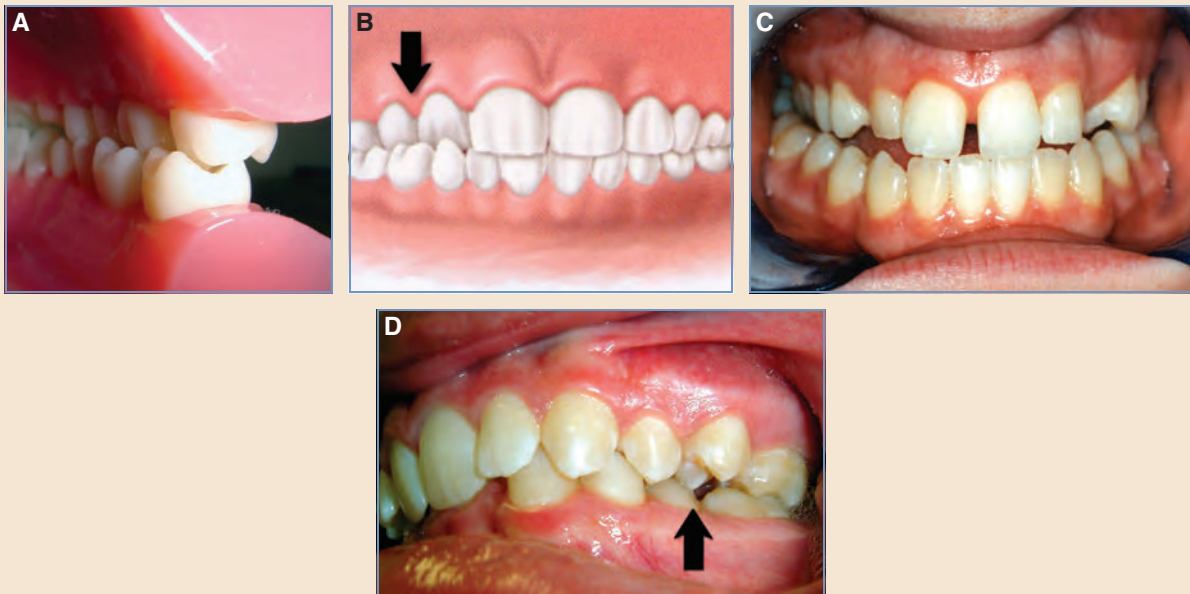


Fig. 29-7 **A**, A model view from inside the mouth demonstrating the lingual cusps of the maxillary teeth occluding the corresponding fossa of the mandibular teeth. **B** and **C**, Lingual crossbite. The buccal cusps of the maxillary teeth are lingual to the buccal cusps of the mandibular teeth. **D**, Buccal crossbite. The maxillary teeth are positioned too far buccally, resulting in no occlusal contact.

If there is excessive overlap of the incisors, this is referred to as a *deep bite* and is measured in millimeters of overlap or as a percentage of tooth crown coverage² (Fig. 29-6).

Transverse dental relationships are difficult to quantify, primarily because there is a large variation in the width of the dental alveolar arch from one person to another. With ideal occlusion, the buccal cusps of the maxillary posterior teeth are positioned buccally to the buccal cusps of the mandibular teeth, and the lingual cusps of the maxillary teeth fit in the corresponding fossa of the mandibular teeth (Fig. 29-7, *A*). Most transverse discrepancies are caused by a narrow maxilla and often result in a unilateral or bilateral posterior lingual crossbite² (Fig. 29-7, *B*). Less frequently, the maxillary teeth are positioned too far buccally, where there is no occlusal contact, resulting in a buccal crossbite (Fig. 29-7, *C*).

DIAGNOSIS AND TREATMENT PLANNING

Before a treatment plan can be formulated, an accurate diagnosis of the patient's problem must be established. A diagnosis of complex maxillofacial problems involving occlusion is based on history, physical examination, radiographic examination, and dental casts that are accurately mounted on an articulator.

Patient History

An accurate understanding of the chief complaint is essential to develop a treatment plan. It is important to obtain a thorough medical and dental history from every patient. The dental specialist should obtain a dental history, noting the frequency of and reasons for dental treatment, along with any previous experience with dental prostheses. The surgeon should also obtain a detailed history from each patient. Diseases such as juvenile rheumatoid arthritis, diabetes, and scleroderma may affect treatment planning. If a patient has a dentofacial anomaly, a history of hyperplasia or hypoplasia resulting from a syndrome, traumatic accident, or neoplasm will affect treatment considerations. In patients with a cleft lip and/or palate, surgical history and any previous diagnostic testing are paramount to develop a treatment plan. A history of previous radiation therapy may limit reconstructive options. The clinician should focus on the history of the particular problem, but it is important to be aware of all aspects of the patient's history to minimize potential complications.

Physical Examination

The focus of the physical examination depends on the patient's particular problem. However, in general, certain aspects are frequently evaluated to a degree for any patient with a craniomaxillofacial problem. The soft tissues should be evaluated for availability, pliability, presence of fistulas, and quality. Some patients with soft tissue deficits may be candidates for autologous reconstruction, whereas others may be better suited for prosthetic reconstruction. Soft tissue may be present, but it may not be of the quality necessary for a particular area of the oral cavity. All of these findings are important for the ultimate rehabilitation of the patient.

The skeletal structures should be evaluated for the quantity, quality, and symmetry of bone. Skeletal deficiencies or defects may be replaced with a corticocancellous bone graft or may require a microvascular flap. Inadequate bone volume in the alveolar ridges may be treated by grafting bone or moving the jaws. Although bone may be present, it may not be of adequate quality to receive an osteointegrated implant or prosthesis. If teeth are present, an effort should be made to preserve the alveolar bone in these areas, and bone grafting should be performed in any areas in which inadequate bone exists.

Occlusion should be evaluated for the presence or absence of each tooth and the level of oral hygiene. Angle's classification of the dentition and jaws as well as any occlusal cant is documented. The midlines of the upper and lower incisors in relation to the facial midline are recorded, and any discrepancies are noted. The normal vertical position of the maxilla results in showing approximately 3 to 4 mm of the upper incisor when the lips are in repose. Any deviations from normal are noted and incorporated into the treatment plan.

Radiographic Examination

Radiographs are important to see any pathologic conditions and evaluate the hard and soft tissues of the facial skeleton for reconstruction. Several radiographic options are available, and each has a specific indication.

Cephalometric Radiographs

Cephalometric radiographs are useful to evaluate the relationships of teeth to underlying bone, the upper and lower jaws to one another, and the jaws to the cranial base (Fig. 29-8). These radiographs are most commonly used for treatment planning of orthognathic surgery. However, they are also useful for posttraumatic reconstruction cases in which a jaw discrepancy exists and the surgeon is uncertain of the etiologic factors. In posttraumatic cases, jaw discrepancies could be the result of a preexisting discrepancy, inadequate reduction of the initial injury, or loss of bone that gives the illusion of a jaw discrepancy. Careful review of the cephalometric films in the context of the patient's history and physical examination findings is very useful to determine the true cause of the problem. Cephalometric films are also useful for treatment planning of surgical reconstruction.

Panoramic Radiographs

Panoramic radiographs provide useful information, such as interproximal bone height, quality of bone in the maxilla and mandible, location of plates and screws, sinus configuration, location of the inferior alveolar canal, anatomy of the mandibular condyles and eminences, impacted teeth, missing teeth, and significant areas of dental decay and periapical disease. This is an inexpensive method to document a postsurgical result with minimal radiation to the patient. Some hospitals do not provide these images, but almost all orthodontists have these machines in their offices. An orthodontist on the team can usually provide these images.

Individual periapical and bitewing radiographs provide superior accuracy and definition to evaluate interproximal alveolar bone height between teeth, caries, ill-fitting restorations, and periapical pathologic findings of potential supporting teeth for fixed and/or removable prostheses. These are the best images to evaluate individual teeth and are taken by a dentist in the office. These images are rarely necessary for a plastic surgeon.

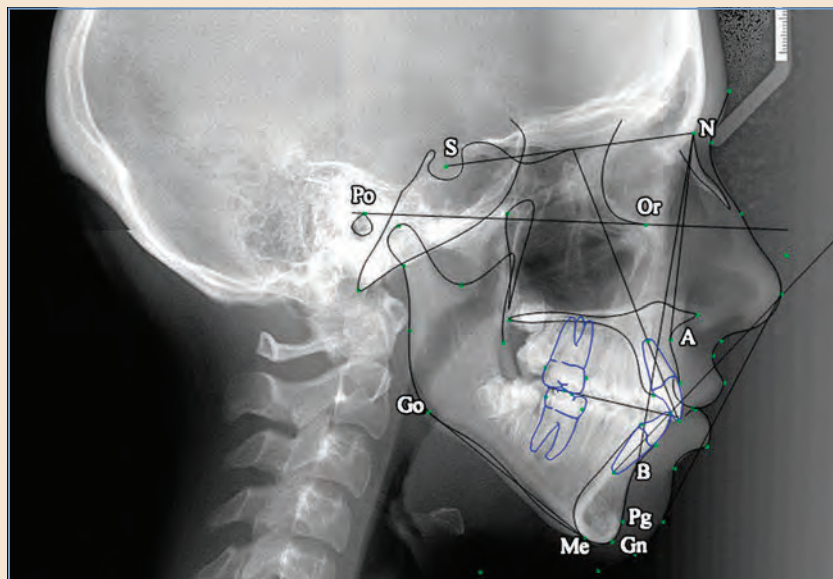


Fig. 29-8 Lateral cephalometric radiograph with common landmarks. (*A*, A point: innermost point in the depth of the concavity of the maxillary alveolar process. *B*, B point: innermost point on the contour of the mandible between the incisor tooth and the bony chin. *Gn*, gnathion: center of the inferior contour of the chin. *Go*, gonion: most inferior and posterior point at the angle, it is formed by the ramus and body of the mandible. *Me*, menton: most inferior point on the mandibular symphysis. *N*, nasion: anterior point of the intersection between the nasal and frontal bones. *Or*, orbitale: lowest point on the inferior margin of the orbit. *Pg*, pogonion: most anterior point on the contour of the chin. *Po*, porion: uppermost lateral point on the roof of the external auditory meatus. *S*, sella turcica [midpoint of the sella turcica].)

DentaScan

An increasingly popular radiographic tool for surgeons and restorative dentists is CT imaging with the DentaScan (GE Healthcare) (Fig. 29-9). The DentaScan images the jaws in thin slices in the coronal plane, giving views of each tooth and the surrounding bone in millimeter increments. These images also provide a cross-section of the jaw from third molar to third molar. Each image is numbered so that any location can be precisely identified. This detailed imaging allows a surgeon to accurately determine (within millimeters) the location of the inferior alveolar nerve and the quantity and quality of bone at any location. The DentaScan has proved to be an accurate method for preimplant assessment (Lacan) and evaluation of bone grafts of alveolar clefts. An area of particular relevance for the DentaScan is preparing osteointegrated implants for posttraumatic edentulous patients.

Successful application and interpretation of radiographic findings allow all members of a reconstructive team to identify and address specific concerns that affect a patient's result.

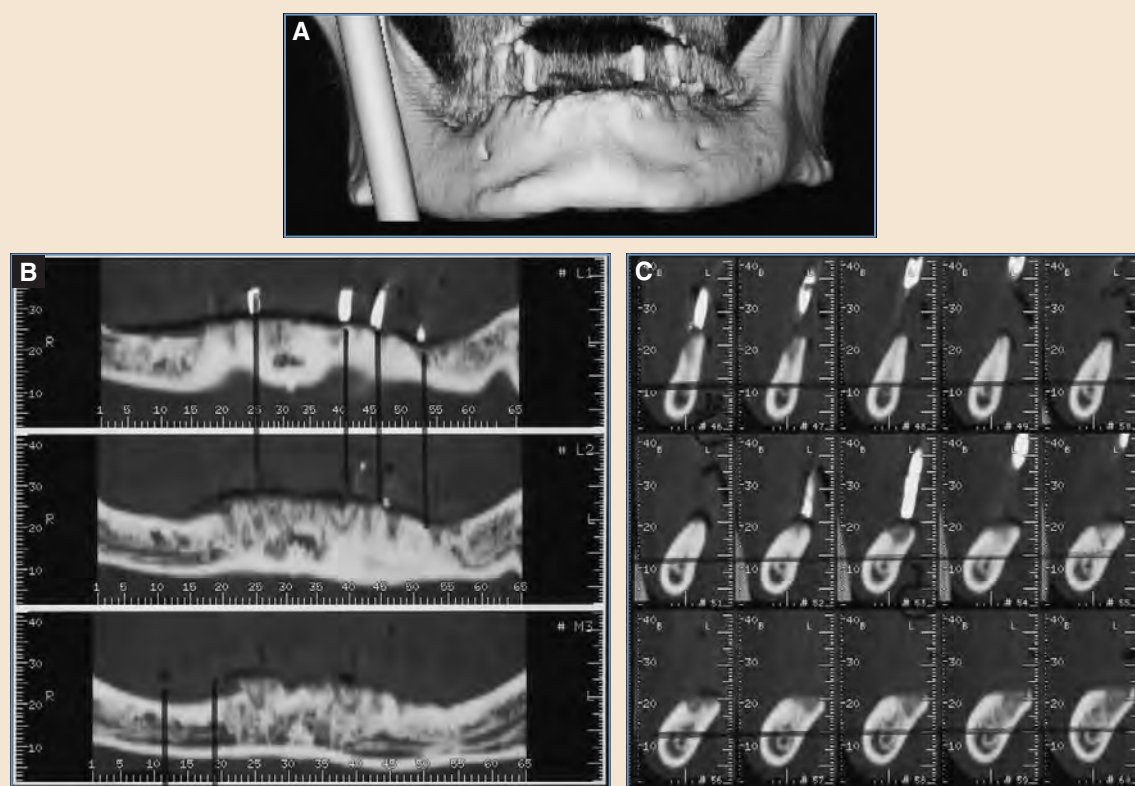


Fig. 29-9 A, The DentaScan is a CT imaging device that allows millimeter measurements of bone in the den-toalveolar region. B, Two-dimensional renderings of the jaws have each representative slice numbered in order. C, Cross-sectional images of the jaws are fabricated and numerically coded to identify the exact location of the image. The quality and quantity of bone in each location can be assessed for placing osteointegrated implants.

Articulation of Dental Casts

Because the extent of tissue abnormalities in oral cavity and jaw relationships is often difficult to see during intraoral inspection, articulated diagnostic casts are essential for planning surgical and prosthetic treatment. These mounted casts provide diagnostic information that may be difficult to obtain from clinical examination alone. *Dental casts* are plaster models of the patient's maxillary and mandibular occlusion that are made by pouring plaster into dental impressions. After the casts have been obtained, they are mounted on an articulator.

An *articulator* is a mechanical instrument, to which maxillary and mandibular casts are attached, that replicates the temporomandibular joints and jaws to simulate mandibular movements. Articulators range in complexity from simple hinge devices to complex instruments that accept three-dimensional dynamic patient registrations. Simple hinge articulators are referred to as nonadjustable, because they are only capable of simple hinge motion, and they do not replicate lateral mandibular movements (Fig. 29-10). Semiadjustable articulators replicate average mandibular movements, such as lateral, protrusive, and *Bennett movements* (lateral shift of the mandibular condyles and articular discs in the direction of the working bite) to varying degrees. Semiadjustable articulators are most suitable for orthognathic surgery, preprosthetic surgery, and maxillofacial prostheses (Fig. 29-11).



Fig. 29-10 The Galetti articulator allows the upper and lower casts to be secured with screws, and the two universal joints allow the clinician to place the casts in various positions. This articulator is useful to place the final splints in orthognathic surgery or to assess dental occlusion as the jaws are moved into different positions.

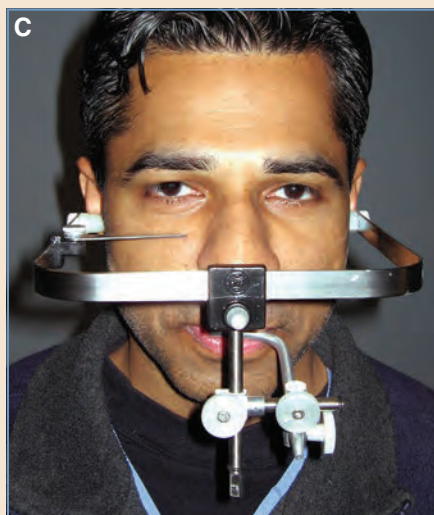


Fig. 29-11 **A**, The facebow and bitefork provide an accurate method of preserving the maxillary-cranial relationship when mounting the maxillary cast. **B**, Aluwax impression wax is heated in warm water and placed on the bitefork of the facebow. The facebow is attached to the fork, placing the earpieces into the external auditory meatus and the inferior orbital rim marker at the rim. The universal screws are tightened. **C**, The facebow is secured to the articulator with the inferior orbital rim marker at the level of the mounting ring. The maxillary cast is placed so that the teeth index into the wax bite on the facebow. The maxillary cast is mounted on the articulator so that its position simulates the craniomaxillary position of the patient. The mandibular cast is mounted by using an occlusal wax registration from the patient. The articulator is turned upside down, and the mandibular cast is related to the maxillary cast with the wax bite registration. **D**, After the plaster is dry, the articulator is turned back to its proper position, and the patient's casts are in proper position to plan the surgery.

When mounting casts on an articulator, it is desirable to orient the maxillary dental cast in the same direction as the opening axis of the articulator. This relationship is preserved by using a facebow. A *facebow transfer* is a caliper device that records the relationship of the maxillary arch to some anatomic reference point or points and transfers this relationship to an articulator. In most instances, the anatomic reference is the transverse horizontal axis between the condyles and inferior orbital rim. The ear opening is a reference point for most semiadjustable facebows, because it is approximately ¼ inch behind the temporomandibular joint. The facebow is positioned with the caliper ends in each ear and the infraorbital pointer at the inferior orbital rim (see Fig. 29-11, C). The universal joints are tightened, and the facebow is removed.

It is secured to the articulator so that it will support the maxillary cast in the proper position for mounting (see Fig. 29-11, A). The teeth of the maxillary cast index into the wax bite pad on the facebow (see Fig. 29-11, B). An arbitrary mounting of the maxillary cast without a facebow transfer can introduce occlusion errors and compromised aesthetics resulting from incorrect orientation of the cast in an anteroposterior and/or transverse direction. Mounting the maxillary cast on the articulator allows the mandibular cast to be positioned with the aid of a patient interocclusal record (see Fig. 29-11, D). The wax record of centric relation and/or maximum intercuspation is used to relate the position of the lower cast to the mounted upper cast. After the lower cast is mounted, the lateral (right, left) and protrusive (optional) records are used to program the condylar guides of the articulator (standard values for Bennett movement is 15 degrees, and condylar guidance is 30 degrees).

The simplicity and speed with which the necessary facebow, dental casts, and interocclusal records are obtained and transferred to a semiadjustable articulator allow an operator to evaluate occlusion and relationship of the jaws with great precision and without the time-consuming techniques needed for fully adjustable articulators.

CEPHALOMETRICS FOR PLANNING ORTHOGNATHIC SURGERY

Although cephalometric treatment planning is most commonly used for traditional orthognathic surgery, it is also indicated for posttraumatic reconstruction, postoncologic reconstruction, and dental rehabilitation. All malocclusions are a result of an improper relationship of the jaws or the position of the teeth within them. *Cephalometric analysis* of radiographs is a quantifiable method of comparing the relationships of the jaws to the cranial base and each other, as well as the relationship of the maxillary and mandibular dentition. This analysis provides information to identify the anatomic basis for malocclusion.

Cephalometric Analysis

Cephalometric analysis begins by identifying a series of standard anatomic landmarks on a radiograph and either tracing them onto a clear acetate overlay or digitizing them on a computer. The landmarks are chosen to include all of the major components of the face (see Fig. 29-8). Lines are drawn between specific landmarks to create a series of planes and angles that enable vertical and horizontal comparisons. Most analyses include a reference area or line for comparison.

Early anthropometric studies on dry skulls used the Frankfort plane, because it closely resembles the natural head position. The Frankfort plane extends from the upper rim of the external auditory meatus (porion) to the inferior border of the orbital rim (orbitale). The Frankfort plane is still commonly used today, but the landmarks are often difficult to accurately identify on cephalometric radiographs. Another reference plane, popularized by Cecil Steiner in the 1950s,

uses a line drawn from the sella turcica (S) to the nasion (N). These landmarks are easily identified on most cephalometric radiographs, making the S-N plane a common reference plane in various cephalometric analyses.³

Extending a line from the nasion (N) to the anterior maxilla (A point) forms the angle SNA. This angle reveals the anteroposterior position of the maxilla relative to the cranial base. Normative data for white Americans indicate that the SNA angle is 82 ± 2 degrees. Therefore a measurement greater than 84 degrees indicates maxillary protrusion. Similarly, angle SNB (line extending from the nasion [N] to the anterior mandible [B point]) reveals the anteroposterior position of the mandible, with normal being 78 ± 2 degrees. An SNB angle of 74 degrees or less indicates mandibular retrognathia, the most common skeletal dysplasia. An SNB angle greater than 80 degrees suggests mandibular prognathism. Angle ANB, the difference between SNA and SNB, is significant, because it indicates the magnitude of the skeletal discrepancy. An ANB angle greater than 2 degrees indicates that the mandible is posterior relative to the maxilla, which is a class II skeletal relationship; it is more likely to appear as a class II dental malocclusion. Conversely, an ANB angle less than 2 degrees demonstrates relative mandibular prognathism and a tendency toward a skeletal class III relationship and a likely dental class III malocclusion.^{3,4}

By identifying multiple dental and skeletal landmarks, a patient's actual dentofacial dimensions are revealed, allowing easy comparison with population norms. However, the clinician needs to be careful not to rely on any single measurement as a sole indicator of maxillomandibular discrepancies, because individual angles are often affected by other angles. For example, angle ANB is greatly affected by vertical proportions. As the mandible rotates in a clockwise direction, for example, in individuals with long faces, the ANB angle increases. This may falsely indicate mandibular deficiency when in fact if the vertical dimension was normal, the counterclockwise rotation of the mandible would likely indicate a normal anteroposterior position. Therefore the cephalometric analysis should be viewed in its entirety when dentofacial dimensions are evaluated. Computers have enabled the input of many more landmarks, allowing clinicians to use multiple analyses to aid in the diagnosis of these discrepancies.

PRESURGICAL ORTHODONTICS

The goal of presurgical orthodontics is to ideally position the teeth to facilitate a planned surgical procedure. One typically associates a need for orthodontic treatment with elective orthognathic surgery. However, there is a role for presurgical orthodontics with patients undergoing post-traumatic reconstruction who have a dentofacial deformity after initial treatment. The purpose of presurgical orthodontics includes aligning the dentition, establishing proper incisor positions, creating a compatible interarch relationship, and preparing the arch before placement of osteo-integrated implants. Attempts to perform orthognathic surgery without orthodontics often lead to poor occlusion, increased risk of relapse, and insufficient surgical correction.

Most malocclusions develop some degree of dental compensation to disguise the inherent discrepancy. Class II malocclusions often present with the mandibular incisors flared anteriorly to lessen the overjet. Excessive overjet also reduces the contact that the mandibular incisors have with the lingual surfaces of the maxillary incisors, causing them to overerupt. This leads to an excessive curve in the mandibular occlusal plane (Fig. 29-12). Conversely, class III malocclusions often present with retroclined mandibular incisors and flared maxillary incisors (see Fig. 29-3). These compensations must be removed before surgery to eliminate dental interferences that may impede adequate bone positioning. Ideally, positioning the incisors in the alveolar bone requires “decompensating” the dentition and leads to a worsening of the occlusion before surgery.



Fig. 29-12 An excessive curve in the mandibular occlusal plane is often the result of overeruption of the mandibular incisors. The mandibular incisors are free to overerupt passively if they do not contact the lingual surfaces of the maxillary incisors.

Establishing compatible dental arches usually involves coordinating the interarch transverse relationships. Because the transverse width of the mandible and maxilla increases posteriorly, any anteroposterior change in the jaws can create dental interferences or a crossbite. Interarch compatibility is evaluated in the later stages of presurgical orthodontics by hand articulating opposing dental casts. This allows an orthodontist to simulate the postsurgical dental relationship and identifies areas that require adjustment. A new set of dental casts is obtained at each subsequent visit to evaluate the adjustments until the desired occlusal relationship is achieved and the dental arches are compatible.

Stabilizing archwires are placed at least 1 month before fabrication of surgical splints to ensure that the dentition will not change before surgery. Surgical hooks are soldered or securely crimped to the stabilizing arch wire as attachments for fixation wiring.

Dentofacial Orthopedics

Jaw discrepancies occur in all three planes of space and are often apparent from an early age. *Dentofacial orthopedics* involves the use of appliances to control and modify facial growth and development to improve these improper skeletal relationships. Traditional growth modification involves the use of intraoral or extraoral forces applied directly to the teeth to produce indirect skeletal forces aimed at improving the maxillomandibular relationship. Common examples include maxillary expansion (Fig. 29-13, *A*), reverse-pull headgear (Fig. 29-13, *B*), and cervical headgear (Fig. 29-13, *C*). These modalities are largely effective and are an accepted part of contemporary orthodontic practice. However, because the forces are directed through the dentition, there are often unwanted dentoalveolar changes. Bone-anchored titanium miniplates offer a method of applying direct bone-borne forces between the maxilla and mandible and have demonstrated tremendous promise, especially for maxillary protraction.^{5,6} This protocol uses Bollard-style plates that are attached to the zygomatic buttress of the maxilla and between the mandibular canines and lateral incisors and have an attachment that perforates at the mucogingival junction (Fig. 29-14, *A* through *K*). Intraoral elastics are worn between the attachments for 22 hours per day and produces a force vector that anteriorly advances the deficient maxilla and soft tissue.^{5,6}

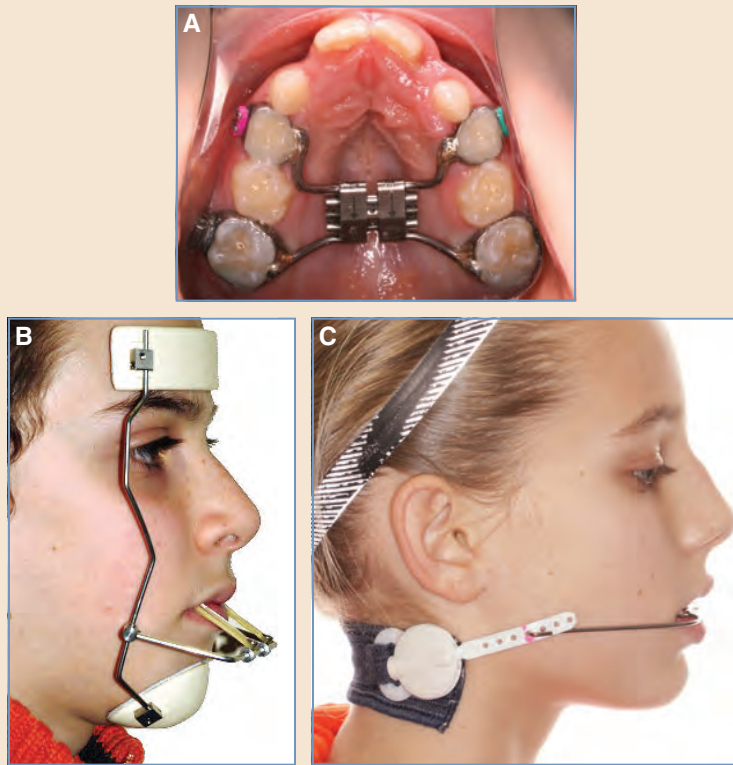


Fig. 29-13 Examples of dentofacial orthopedics. **A**, Maxillary expansion. **B**, Reverse-pull headgear. **C**, Cervical headgear.



Fig. 29-14 A-C, Profile and intraoral pretreatment photos demonstrating deficient maxilla and negative dental overjet in an 11-year-old boy.

Continued



Fig. 29-14, cont'd D, Bone-anchored titanium miniplates placed in the maxilla. E, Bone-anchored titanium miniplates placed in the mandible. F-H, Profile and intraoral photos after 26 months of interarch elastics and minimal orthodontics. Profile is notably more balanced, and dental overjet is now positive. I-K, Posttreatment photos after comprehensive orthodontic treatment at age 15 years. Profile remains balanced and dental occlusion is ideal.

Osteointegrated Implants

Osteointegrated implants may be the treatment of choice for skeletally mature patients in need of prosthetic tooth replacement. These patients have adjacent teeth with a history of minimal restorative treatment or none at all and bone sites that allow an implant of optimal length, diameter, position, and angulation. To properly place osteointegrated dental implants, one needs to know the proper location of the teeth. In an edentulous patient, it is difficult to see exactly where the teeth will be. The prosthodontist should fabricate a diagnostic waxup of the occlusion to help make this determination. A waxup consists of recreating the missing teeth from wax on articulated plaster casts. After the teeth have been waxed into the proper position, the desired location of the implants can be determined. The implant locations are marked on the plaster cast, and a stent is made that incorporates radiopaque markers at the desired location of each dental implant. The patient wears this stent while undergoing DentaScan imaging so that the proposed location of each implant can be seen on the images. Because the markers are at the implant site, the clinician can determine whether the bone in each location is of sufficient quantity and quality. Also, the maximal implant length can be determined on the basis of measurements obtained from the image. If the desired location of the implant is compromised, an alternate site is selected, or augmentation bone grafting is performed (see Fig. 29-9).

Implant dimensions can be determined by bone mapping, periapical radiographs, CT scans, and dental casts. Implants should be surrounded by 2 mm of bone to prevent outward bone resorption, ensure implant stability and correct labiopalatal implant placement, reduce occlusal stresses to the bone, and allow formation of an acceptable papilla between implant and tooth. The implant platform should be situated 2 mm palatal to the predetermined incisal edge of the final restoration. The restoration can be screw retained if the access hole is lingual to the incisal edge, which allows crown retrievability and aesthetic labial contouring of the crown. At least 2 mm of keratinized gingiva should be present on the crest so that it is also present on the abutment and crown.⁷ Coronoapical placement of the implant is critical to provide a smooth emergence profile of the definitive restoration that is similar to the natural tooth at the same level and gives the appearance of a root prominence.⁸ The thickness of the periodontal type determines the length from the implant to the free gingival margin of the crown. The platform or head of the implant should ideally be 2.5 (thick periodontal types) to 3 mm (thin periodontal types) apical from the desired final labial gingival margin of the definitive crown. Adequate space must exist between the implant platform and opposing dentition for anatomically correct restorations. The interocclusal space between the opposing dentition and top of the implant platform should be at least 6 to 7 mm (that is, 2 mm metal and ceramic, 4 mm abutment height, and 0.5 mm for the abutment collar).⁷

Ideal periimplant aesthetics require preservation of intact underlying bone and adjacent teeth; however, interproximal bone loss is inevitable after tooth extraction, which causes apical migration of the interdental papilla. Implant clinical crowns are usually longer than the original tooth-to-tooth contacts to compensate for the lack of interdental papilla. To avoid technique-sensitive grafting procedures that may be necessary in the presence of inadequate hard and soft tissues at healed extraction sites, current tissue preservation techniques involve immediate replacement of failing teeth (without acute infection or bone destruction) with provisional implant restoration. Studies have shown that immobile immediate provisional restorations can enhance soft tissue management by supporting the periimplant mucosa and maintaining papilla height and gingival outline. Delayed implant placement requires several months of treatment but can produce equivalent results if the bone support is intact, length of treatment is reduced, and number of surgical procedures is reduced. The overall success rate for immediate implant placement is 94.6% compared with 94.4% for implants placed at healed sites.⁹

Research over the past decade has helped to predict achievable papilla height between two teeth, between teeth and implants, and between adjacent implants in the maxillary anterior segment. It is recommended that the clinician provide an adequate mesiodistal distance of at least 1.5 to 2 mm between an implant and adjacent tooth and at least 3 to 3.5 mm between two implants to prevent interimplant crestal bone loss resulting from the crossover of angular defects.¹⁰ If the two maxillary central incisors are replaced with two implants, a 4 to 4.5 mm distance is recommended.¹¹ Interdental papilla completely extending up to the contact point between two natural teeth is directly related to the distance between the contact point and the interdental crest of bone. It has been reported that the papilla is present 100% of the time if the distance between two teeth is 5 mm or less; however, the papilla was present 56% of the time at a distance of 6 mm.⁸ Papilla reform predictably between an implant and a natural tooth when the distance from the crestal bone to the contact point is 4.5 and 3.5 mm between adjacent implant restorations. The greatest vertical soft tissue height can be achieved between a tooth and a pontic (6.5 mm) and between adjacent pontics (6.0 mm) without proximity limitations.¹² These distances should be considered when placement of implants and/or pontics is determined during restoration of single or multiple missing teeth in the aesthetic zone. Implants with smaller diameters at the implant-abutment interface or fewer implants with pontics may be necessary to maintain the papilla and improve the aesthetics of multiple missing teeth in the aesthetic zone.

Prosthetic Replacement of Missing Teeth

Patients with clefts of the alveolar processes and palate may have missing teeth (that is, lateral incisors) and deficient soft and hard tissue requiring surgical reconstruction. It is possible to replace a single tooth in the aesthetic zone with a removable (interim/transitional) partial denture, a fixed partial denture/bridge, or an implant-supported crown. When choosing the type of prosthesis, the following factors must be taken into consideration: patient age, skeletal growth, occlusion, periodontal status, tooth eruption patterns, orthodontic treatment, quality and quantity of hard and soft tissue, oral hygiene, psychological state, and patient preference for removable or fixed prostheses. Osteointegrated implants are usually the treatment of choice for skeletally mature patients when anterior teeth neighboring the edentulous space have had minimal or no restorative treatment. A removable appliance is recommended for pediatric patients with tooth loss, because the prosthesis can be adapted and adjusted easily according to growth, dental changes, and bone grafting procedures to the edentulous area. Secondary bone grafting to support or augment maxillary segments is beneficial for definitive prosthetic treatment during early adulthood, because it allows a reduction in the length of fixed partial dentures (fewer abutments) and provides adequate quantity and quality of bone when the prosthodontist is considering placement of an osteointegrated implant. To deliver an aesthetic restoration and provide an optimal occlusal scheme for a patient, an orthodontist should be consulted to ensure the completion of maxillary and mandibular growth and orthodontics before the fabrication of definitive fixed partial dentures/bridges and implant-supported restorations.

Resin-bonded fixed partial dentures for single tooth replacement are a minimally invasive treatment option for patients in their late teens or in adulthood who may opt not to have implants or conventional fixed partial dentures, those who are not candidates for implant-supported restorations (because of incomplete skeletal growth, lack of hard/soft tissue, medical or psychological issues, or local adverse conditions), or those who desire an aesthetic fixed restoration requiring little or no preparation of neighboring teeth before additional bone grafting and/or implants. Composite and ceramic resin-bonded fixed partial dentures provide superior aesthetics for traditional cast metal resin-bonded fixed partial dentures, or “Maryland bridges,” because of the absence of cast metal that often causes discoloration of abutment teeth in translucent areas.

CASE EXAMPLES

The following two patients demonstrate how the preceding concepts were used to manage complex cases that involved occlusion.

The first patient was a young woman who sustained major maxillofacial trauma that included injuries to her mandible, maxilla, and bilateral orbit and malar regions. The maxillary injury included loss of her anterior teeth and a major avulsion of alveolar bone. The plain radiograph showed the location of her plates after fixation (Fig. 29-15, *A*). Because the patient had lost anterior maxillary bone, it was uncertain whether the apparent class III malocclusion was the result of bone loss or skeletal malocclusion. Articulated models were used with cephalometric analysis to determine whether the maxillomandibular relationship was normal and the cause of the deficient maxilla was the result of avulsed bone.

The malunion of the mandible required treatment, and the articulated models were used to perform model surgery in preparation for the mandibular osteotomy (Fig. 29-15, *B*). A temporary maxillary prosthesis was also fabricated by the prosthodontist (Fig. 29-15, *C*). The mandible was moved into the ideal position and secured with rigid fixation (Fig. 29-15, *D*). Because osteointegrated implants were to be placed, an orthodontist aligned the arches before implant placement (Fig. 29-15, *E*).



Fig. 29-15 *A*, Postoperative radiograph of the patient after panfacial fracture. *B*, Facebow-mounted dental records showed an inadequate reduction of the mandibular fracture. These records were also used by the orthodontist to determine how much orthodontic movement was needed to achieve class I occlusion. *C*, Postmodel surgery records with temporary dental prosthesis in place.



A DentaScan was used to determine the amount and location of bone to be grafted (Fig. 29-15, *F* through *H*). Corticocancellous bone from the iliac crest was used to augment the anterior maxilla and nasal floor in preparation for the implants (Fig. 29-15, *I*). A vestibuloplasty with a split-thickness skin graft was performed to extend the buccal vestibule and improve the soft tissue in the region of the anticipated implant abutments (Fig. 29-15, *J*).

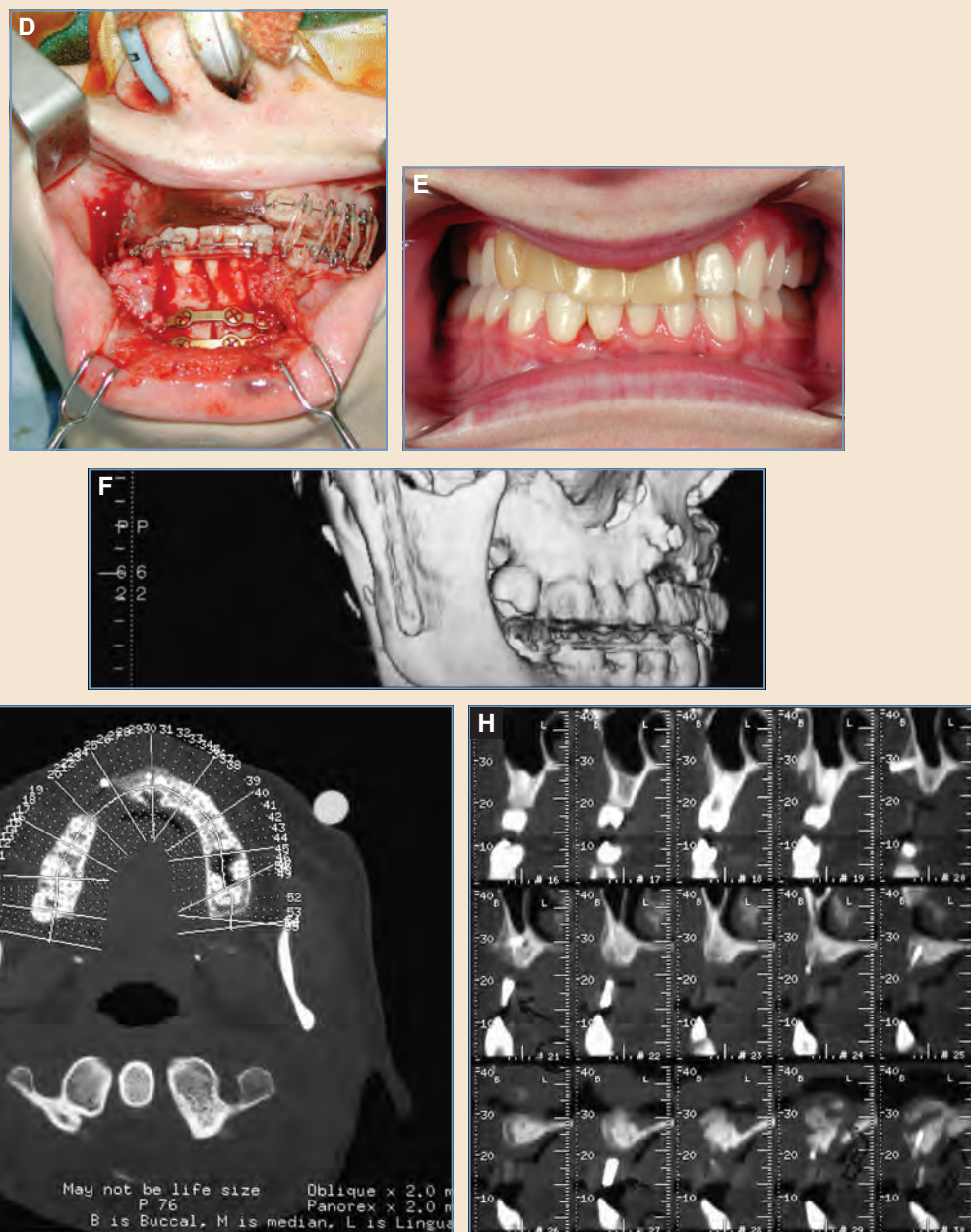


Fig. 29-15, cont'd **D**, Postreduction view after posttraumatic orthognathic surgery. **E**, Patient as she appeared during the extended treatment phase. **F**, Patient's DentaScan and records showed that her class III appearance was the result of avulsive bone loss at the anterior maxilla rather than incorrect reduction of fractures. **G** and **H**, DentaScan images showed the maxillary dental arch with numbered images. For this patient, imaging was done with a surgical stent containing radiopaque markers at the planned location of each implant. The markers allowed the surgeon to key in on the exact numbered image to assess bone quality and density.

The implants were ready to be placed 6 months after the bone grafting. The desired implant location was determined from the temporary prosthesis. It is important to position the implants in the center of the desired location of the tooth to avoid compromising dental aesthetics by having an implant that is visible between the teeth. A surgical stent was fabricated to indicate the exact location of each implant during surgery (Fig. 29-15, *K* through *L*). The implants were placed in a minimally traumatic fashion under copious irrigation (Fig. 29-15, *M*). As each implant was placed, a pin was placed to facilitate parallel positioning of all of the implants (Fig. 29-15, *N* and *O*).

The final periapical radiograph of the implant showed good bone apposition with minimal resorption of bone (Fig. 29-15, *P*). Finally, the patient's smile can be appreciated after final cementing of the prosthesis onto the implants (Fig. 29-15, *Q*).

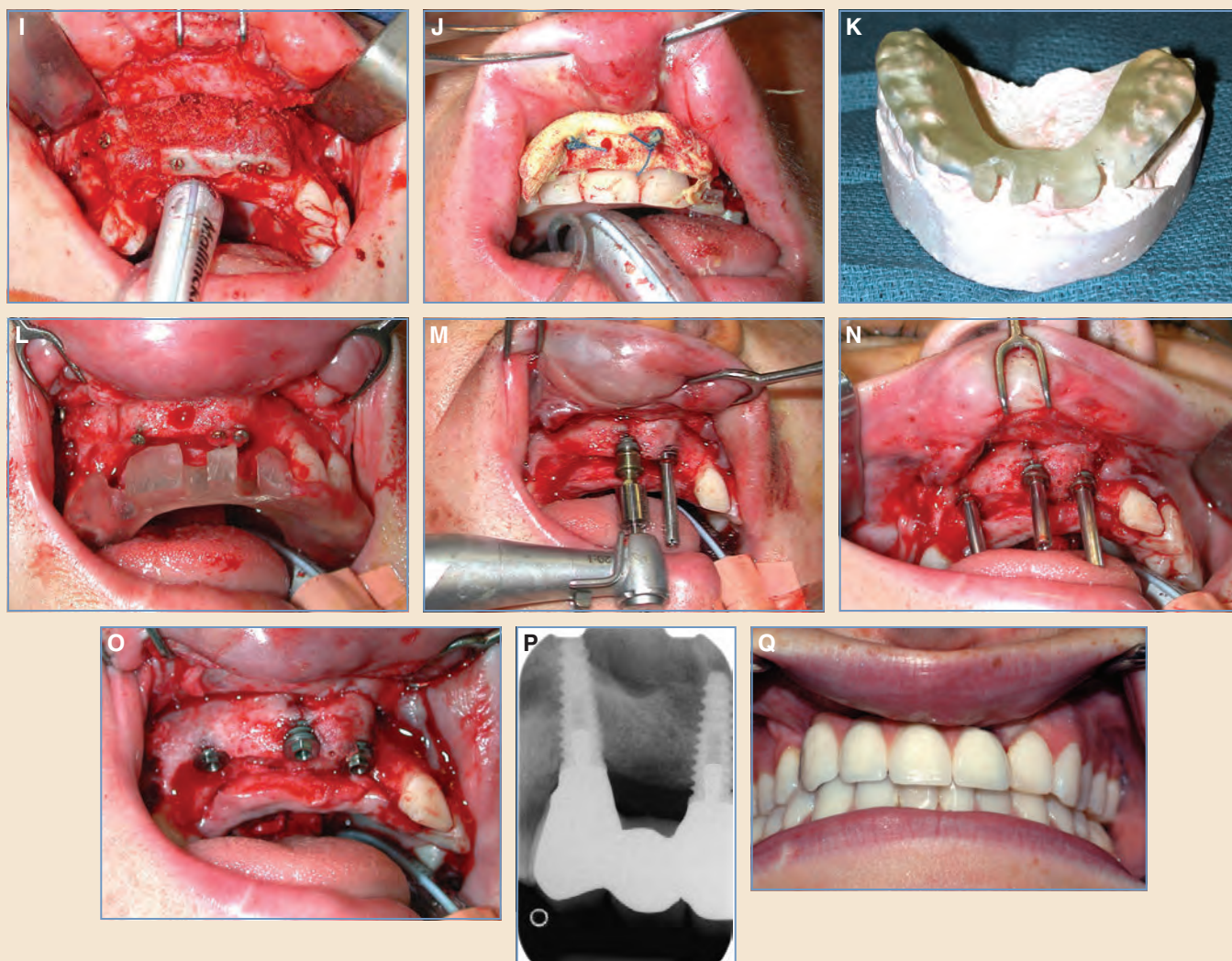


Fig. 29-15, cont'd **I**, Corticocancellous iliac crest bone was placed in the maxilla to augment the osseous volume required for osseointegrated implants. **J**, A skin graft was used to re-create the normal vestibular depth after the bone grafts were placed. **K**, Surgical stent fabricated by the prosthodontist to show the surgeon the exact location and angulation of each implant. **L**, The stent in place showing where the implants will be placed. **M**, The implants were placed with a low-speed drill and irrigation. **N**, Implants showing parallel placement. **O**, Implants at the second stage before transmucosal abutments were placed. Note the excellent take of the grafted bone. **P**, Final periapical radiograph showing the implants. **Q**, Patient's final occlusion after cementation of the final prosthesis.

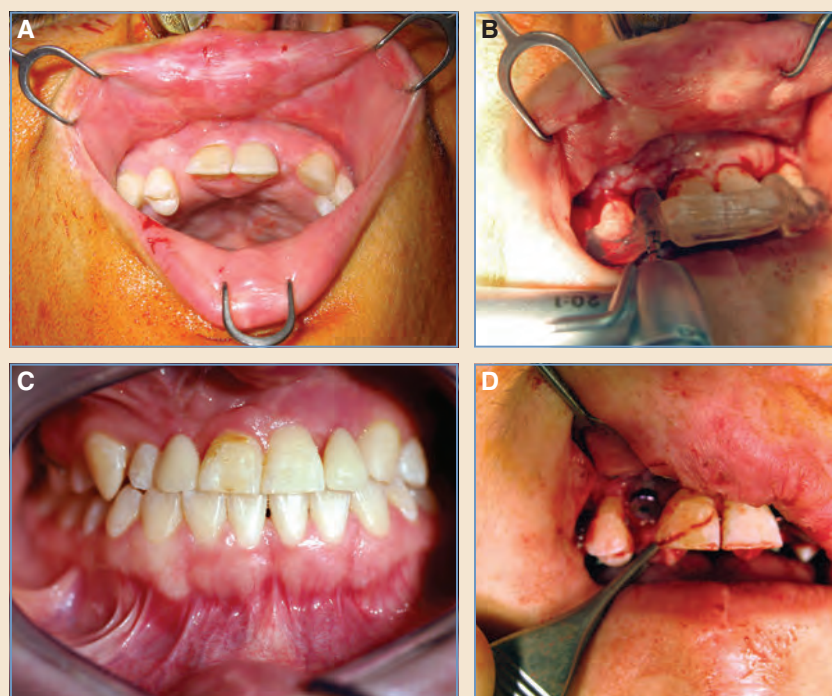


Fig. 29-16 A, This young, partially edentulous woman had undergone bilateral cleft lip and palate repair and desired osteointegrated implants. B, The prosthodontist's surgical stent was used by the surgeon to ensure placement of the implant in the desired position. C, Temporary prosthesis that the patient wore while the implants were undergoing osteointegration. D, Second-stage application of the transmucosal posts to which the final prosthesis was secured.

This 22-year-old woman had undergone a bilateral cleft lip and palate repair and was seen for prosthetic reconstruction of her upper lateral incisors (Fig. 29-16, A). DentaScan imaging revealed insufficient bone for dental implants, so she underwent supplemental alveolar bone grafting 6 months before the implants were placed. Her dental casts were articulated, and the ideal form of the lateral incisors was created from wax on the maxillary cast. From the diagnostic waxup, a surgical stent was fabricated from light-cured acrylic (Triad System; Dentsply). A crestal gingival incision was reflected with a No. 9 periosteal elevator, and a surgical stent was placed in the mouth to show the surgeon the ideal location and inclination of the osteointegrated implants (Fig. 29-16, B). The implants were placed according to the manufacturer's protocol (Southern Implants), and the temporary prosthesis was used for 6 months while the implants osteointegrated (Fig. 29-16, C). The second stage required placing the abutments, which are transgingival posts that screw into the implants and secure the final prosthesis (Fig. 29-16, D).

PROSTHETIC CONSIDERATIONS FOR PEDIATRIC ONCOLOGY PATIENTS

Surgical considerations, such as preservation of the hard palate where possible, retention of key teeth, incision location and resections relative to specific teeth, use of split-thickness grafts, and elimination of tissues that will be rendered nonfunctional postoperatively, should be discussed before surgery to enhance the prosthetic prognosis without compromising tumor removal. It is

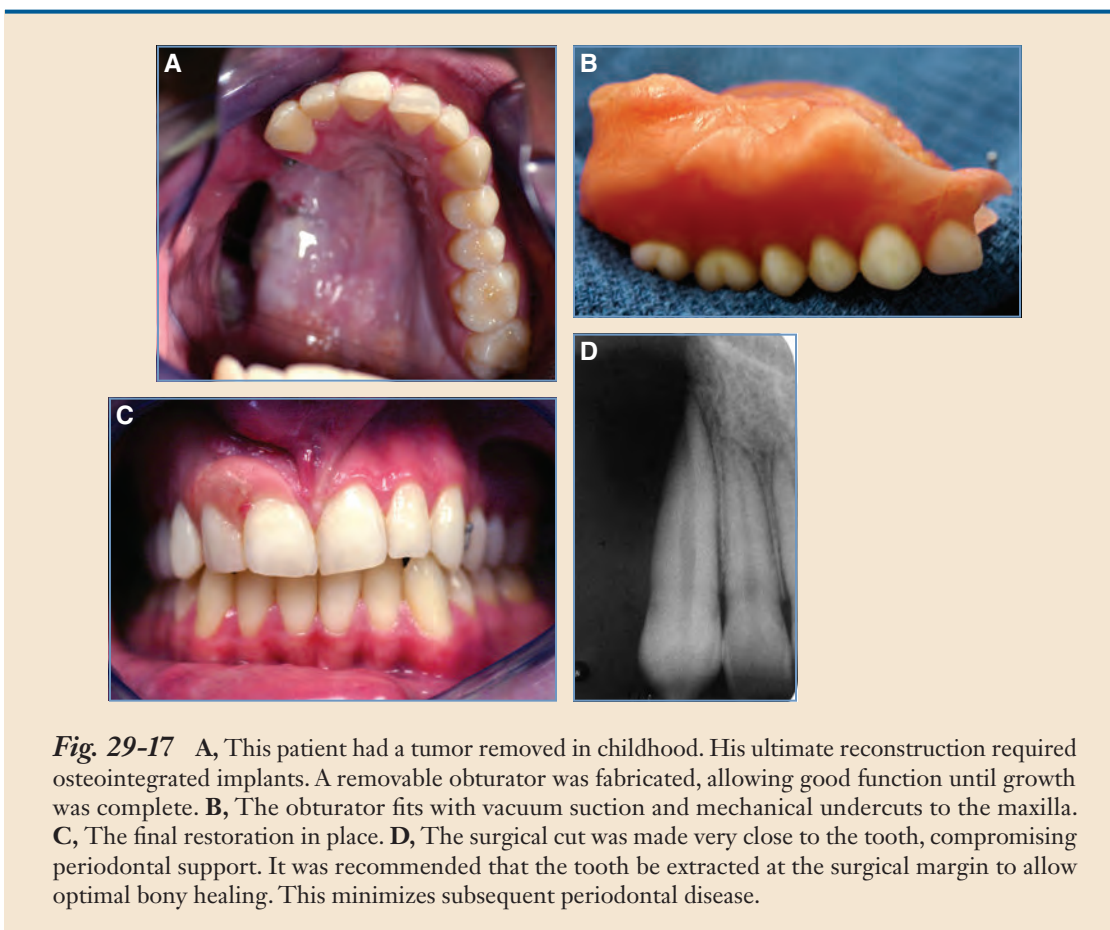


Fig. 29-17 A, This patient had a tumor removed in childhood. His ultimate reconstruction required osteointegrated implants. A removable obturator was fabricated, allowing good function until growth was complete. B, The obturator fits with vacuum suction and mechanical undercuts to the maxilla. C, The final restoration in place. D, The surgical cut was made very close to the tooth, compromising periodontal support. It was recommended that the tooth be extracted at the surgical margin to allow optimal bony healing. This minimizes subsequent periodontal disease.

important to preserve the hard palate and alveolar processes, because this allows support of the prosthesis and possible fabrication of an implant-supported restoration (Fig. 29-17). Maintaining the premaxillary segment is critical for appearance (that is, dental aesthetics and lip support), and this segment is the most effective site for implant placement for edentulous patients. Removable maxillofacial prostheses with attachments on the tissue side may be fabricated to seat over a bar splinting multiple implants or individual implants. The preservation of the arch form allows greater anteroposterior spread of implants, which contributes to greater retention of the obturator prosthesis during speech and function.

Obturation of palatal defects is enhanced when there is healthy periodontal support and multiple teeth are present on both sides of the arch to provide cross-arch stabilization for interim and definitive obturator prostheses. In many patients, the tissue is compromised because of the location and extent of the resection; however, the surgeon should take into account clasping of key teeth such as the cuspid and bony support of the hard palate to provide greater retention, support, and stability of the prosthesis (Fig. 29-17, A through C). Healthy periodontal support of the cuspid and its location at the corner of the maxillary arch are important for retention of a prosthesis and lip support. The lateral incisor is a poor tooth to clasp because of its smaller root and lack of bony support, which may lead to greater mobility of the tooth if it is used to retain a prosthesis. The surgeon should be mindful of the location of the transalveolar resection, because resections through the transseptal bone between teeth have a negative impact on the bony support, mobility, and vitality of the remaining teeth adjacent to the resection (Fig. 29-17, D).

To preserve bony support of the teeth, resections should be made after extraction of the next distal tooth, as far as possible from the tooth adjacent to the defect and through the distal portion of the socket.¹³

The surgeon can contribute to additional forms of obturator retention and tolerance by lining a split-thickness skin graft to the internal cheek surface of the defect and linings within the defect to support modest applications of pressure.¹⁴ Obturator prostheses should engage the scar band superiorly and inferiorly to enhance stability, support, and retention. A split-thickness graft may also be considered to cover the medial margin of the palatal bones when there is inadequate palatal mucosa.¹⁵ Engaging the medial wall of the defect can increase retention of the obturator, which is particularly important when remaining teeth are compromised or alternative forms of retention are inadequate.

The surgeon should remove the entire soft palate on the resected side if less than 1 cm of the posterior one third remains and the patient has teeth remaining for adequate retention and support of a prosthesis. The nonfunctional band of soft palate will contract superiorly, making it difficult to seat or remove the prosthesis, and adequate velopharyngeal closure will be difficult. Speech may be hypernasal, and leakage of foods and liquids into the nose may occur during swallowing if the soft palate remnant prevents optimal placement of the prosthesis for restoring palate-pharyngeal function.¹⁶

Obturator Prostheses

Obturator prostheses are indicated for patients with congenital or acquired tissue openings, primarily of the hard palate and/or contiguous alveolar structures¹⁷ (Fig. 29-18). Obturator prostheses may require an additional speech aid component to prosthetically manage velopharyngeal insufficiency resulting from defects of the hard and soft palates. An obturating prosthesis can be a removable partial denture, a complete denture, an overdenture, or an acrylic resin stent without tooth replacement. The objective of prosthetically restoring palatal defects is to provide the pa-



Fig. 29-18 A, This patient had a large anterior hard palatal fistula that had failed local flap repair once previously. He had not undergone maxillary expansion. B, The cast shows the large fistula just posterior to the incisive foramen. C, The team prosthodontist fabricated a palatal expander that also served as a fistula obturator. This allowed the patient to function normally while the maxilla was expanded. Definitive fistula repair was undertaken after maxillary expansion.

tient with a prosthesis that is comfortable, reestablishes the oral-nasal partition, allows control of resonance and nasal emissions during speech, prevents leakage of foods and liquids into the paranasal sinuses, restores mastication, provides support for the lip and cheek, and is acceptable in appearance. This is especially relevant for providing children with normal function and appearance when surgery needs to be delayed for reasons such as completion of chemotherapy, cessation of growth, or postponing major surgery until the summer. A pressure-resistant seal of the interim and/or definitive obturator bulb against the mucosal lining and skin graft provides physical separation between the oral and sinus cavities necessary to restore speech and swallowing. Normal or near-normal levels of function and aesthetics can be achieved for most patients, with hard palate defects restored with definitive prosthodontic treatment.^{18,19}

There are three phases of prosthetic rehabilitation: surgical, interim, and definitive obturation. The first phase, *surgical obturation*, requires placing the prosthesis at the time of surgery to maintain oral function during the initial postoperative period. The prosthesis is fabricated on a cast made from a preoperative impression. It is possible to retain the prosthesis with either wrought wire clasps or ligature wires. For completely edentulous patients, ligature wires are threaded through the alveolus after penetration of the alveolus with an awl, drill, or 18-gauge needle tap corresponding to the wire openings in the prosthesis. The wires are tightened completely with a clockwise rotation, cut, and twisted interproximally for dentulous patients or twisted back into the wire openings/recesses for edentulous patients. The immediate surgical prosthesis is used for 7 to 10 days, helps the healing process, and prevents scar contraction.²⁰

An *interim obturator* provides patients with a comfortable and functional prosthesis until healing is complete. This prosthesis is most commonly made on a cast from a preoperative impression and is available for insertion when the immediate obturator and surgical dressing are removed. Delivery of an interim prosthesis provides physiologic, hygienic, and psychological support for patients. An impression can be made when the surgical obturator and surgical dressing are removed, but this may delay insertion of the interim obturator and cause increased anxiety because of cosmetic and functional deficiencies. Lack of early obturation of the defect may lead to collapse of tissue around the defect, which may compromise facial aesthetics and complicate future rehabilitative care. An interim obturator is commonly worn for 3 to 4 months for dentulous patients and for 6 months for edentulous patients until healing is sufficient to allow fabrication of a definitive obturator. Additional factors must be considered before fabrication of a more permanent restoration, such as the patient's tissue response to radiation and/or chemotherapy, the size of the defect, progress of healing (soft/hard tissues), presence or absence of teeth, potential for future growth before placement of osteointegrated implants, and effectiveness of the present obturator.

A definitive obturator is indicated when the hard and soft tissues show clinical signs of health and appear stable postoperatively and should only be considered when craniofacial growth has ceased. Definitive obturator prostheses for partially edentulous patients (Fig. 29-19) are similar in construction to definitive removable partial dentures. A definitive obturator provides prosthetic tooth replacement and consists of denture acrylic that is heat processed to a rigid cast metal framework with clasps and cast fittings that fit into prepared surfaces of the teeth for selected teeth to retain and support the prosthesis. An existing denture can be rebased or relined postoperatively, or a new denture/obturator may be fabricated for edentulous patients (Fig. 29-20). A patient's ability to function with (that is, speak, swallow, and masticate) and tolerate the existing interim obturator may provide an indication of prosthetic outcome with a definitive prosthesis; however, the clinician must take into account the prosthetic limitations of an interim prosthesis.

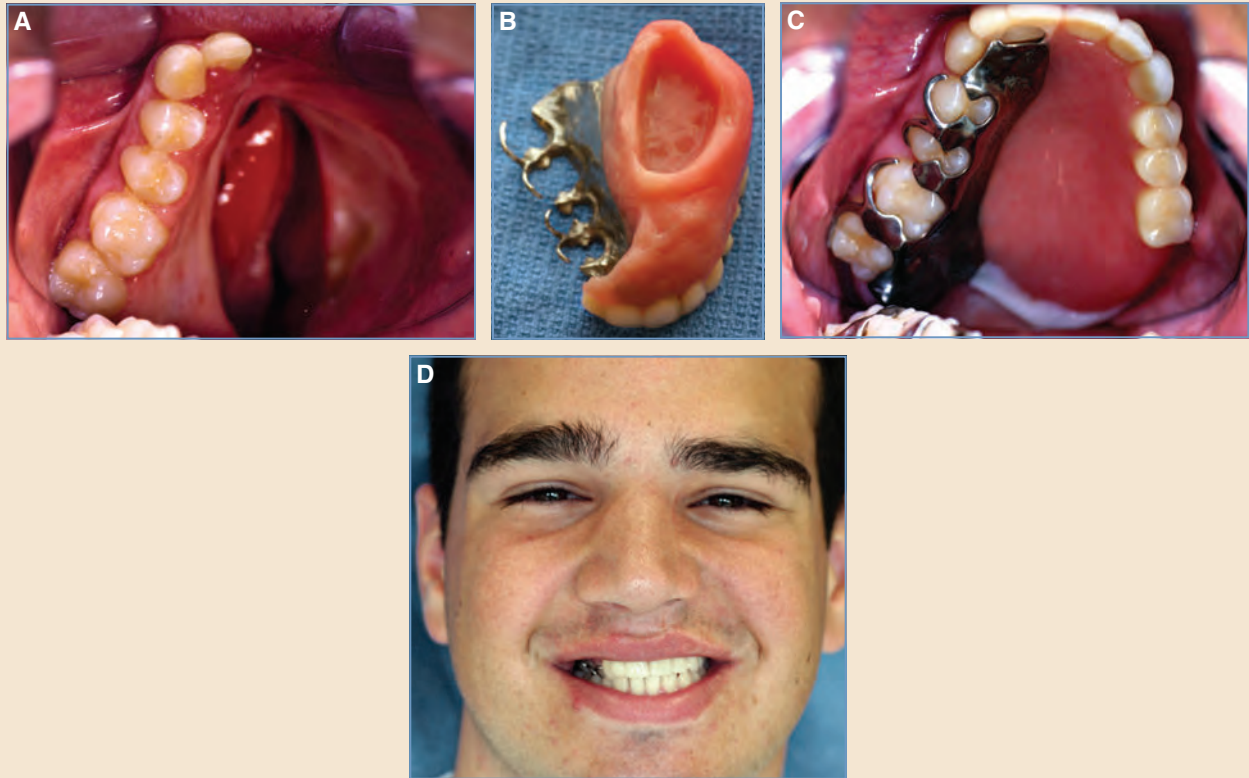


Fig. 29-19 A, Large postoncologic defect in the maxilla of a teenage patient. The growing patient could not have implants placed, so a prosthesis was fabricated to provide normal form and function. B, Prosthesis showing mechanical undercuts and clasps. C, Prosthesis in the maxilla. D, Patient's final occlusion.

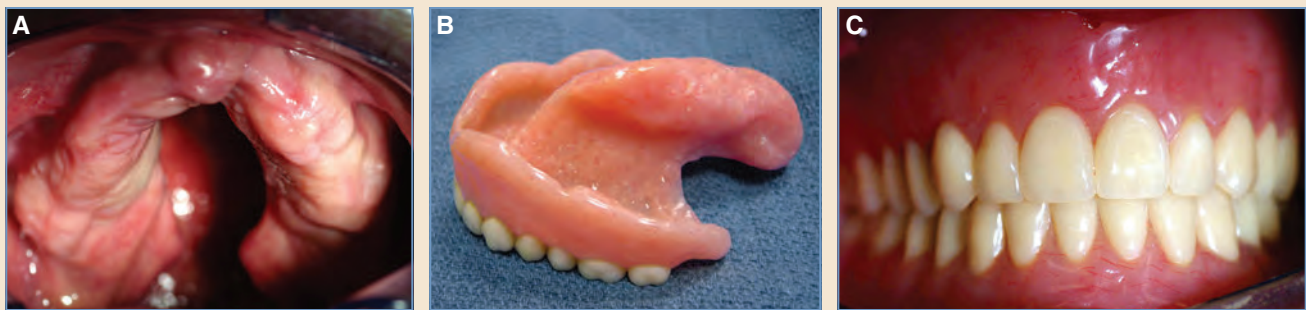


Fig. 29-20 A, This adult patient had previously refused to undergo cleft palate repair but subsequently presented for treatment. B, The prosthesis was designed to obturate the cleft palate and use its undercuts to provide retention. C, Patient's occlusion with the prosthesis in place.

Speech Aid Prostheses

Speech aid prostheses (*soft palate obturators*) are maxillary, removable prostheses indicated for restoring acquired or congenital/developmental aberrations (Fig. 29-21) of the soft palate, with the posterior portion extending into the pharynx to separate the oropharynx and nasopharynx during phonation and deglutition, thereby completing the palatopharyngeal sphincter.¹⁷ Speech aid prostheses are indicated for selected patients diagnosed with velopharyngeal insufficiency (see Chapter 23). It is a condition in which there is a lack of effective closure between the soft palate and one or more of the pharyngeal walls during swallowing or speech sounds that require high intraoral pressure. Patients present clinically with nasal reflux, escape of air during speech, hypernasality, and compromised swallowing. The lack of closure may be the result of palatopharyngeal insufficiency, incompetence (inadequate elevation of soft palate movement), or lack of pharyngeal wall movement.

A speech aid prosthesis helps patients to eliminate nasality, nasal emissions into the defect, and regurgitation of food into the nasopharynx. The prosthetic bulb extends to the posterior pharyngeal wall at the level of the palatal plane and median tubercle of C1 and is functionally developed and modified according to the confines of the muscles of the lateral and posterior walls.²¹ Evalu-

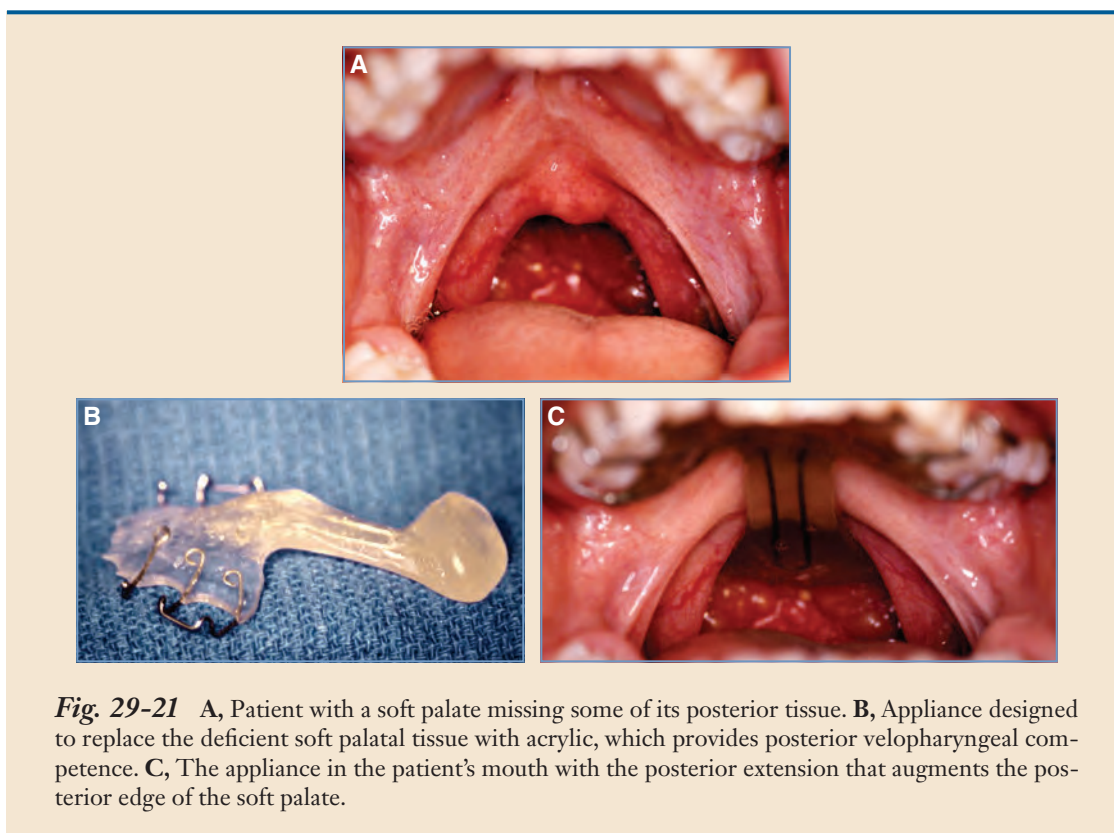


Fig. 29-21 A, Patient with a soft palate missing some of its posterior tissue. B, Appliance designed to replace the deficient soft palatal tissue with acrylic, which provides posterior velopharyngeal competence. C, The appliance in the patient's mouth with the posterior extension that augments the posterior edge of the soft palate.

ation by a speech pathologist is recommended for patients in whom velopharyngeal function is restored, but speech articulation is poor. Speech therapy is extremely important for patients with a cleft palate and compensatory speech patterns, because speech aid prostheses do not eliminate these habits affecting speech. The prosthodontist should consider videofluoroscopy and nasal and oral endoscopy when a prosthesis is fabricated to ensure adequate palatopharyngeal closure for patients presenting with palatopharyngeal insufficiency or incompetence.²²

Palatal Lift Prostheses

Palatal lift prostheses are removable prostheses that can be used as temporary or definitive treatment for palatal incompetence (Fig. 29-22). Velopharyngeal closure is achieved by prosthetic elevation of an incompetent soft palate that is dysfunctional as a result of clefting, surgery, trauma, stroke, or paralysis from an unknown cause. Postoperative scarring after palatoplasty, traumatic brain injuries, cerebral palsy, and injury to the soft palate may cause palatal incompetence. A palatal lift prosthesis differs from a speech aid prosthesis, because it does not extend into the pharynx to restore palatopharyngeal function. The palatal lift elevates the soft palate (adequate length and size) both superiorly and posteriorly to achieve soft palate closure with the peripheral pharyngeal tissues, thereby improving voice quality by increasing oral pressure for consonant articulation and reducing hypernasality and escape of nasal air. Prosthetic stimulation of the soft palate muscles is recommended as soon as palatal paralysis is noted to prevent palatal disuse atrophy. The prosthetic–physical therapy may progress to the point where adequate elevation of the soft palate can be achieved without the prosthesis.²³

Completion of a palatal lift prosthesis may require numerous visits over several months to provide successive additions to the prosthesis to offset tissue tolerance problems on the soft palate and allow the patient to adjust to wearing the prosthesis. It is recommended that the prosthodontist and speech pathologist work together to fabricate the prosthesis to achieve optimal rates of nasal airflow and intraoral air pressures during speech production. Although some clinicians use palatal lift prostheses with the rationale that the palatopharyngeal port function will improve over time as the prosthesis is worn, it appears unwarranted to routinely expect permanently im-

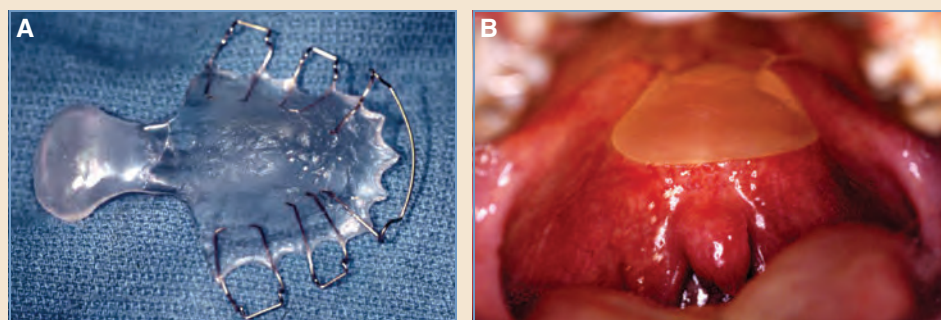


Fig. 29-22 A, A palatal lift, which elevates the posterior portion of the soft palate if incompetence is observed. It can be frequently adjusted or reduced, allowing the patient to achieve normal resonance. B, The appliance can be seen extending back under the soft palate where it elevates this structure and helps it to achieve normal function.

proved palatopharyngeal function during speech production to the point that the prosthesis may be discontinued.²⁴

Commissure Splint Therapy

Treating traumatic injuries to the commissure, such as injuries resulting from electrical or thermal burns, is a challenge for both plastic surgeons and dentists. *Commissure splint therapy*, in association with surgery, has been effective to reduce the effects of scar contracture from oral and perioral burns that can result in cosmetic deformities of the commissure, microstomia, obliterated vestibules, tongue ankylosis, and compromised mastication and speech. Dental deformities associated with contracture have also been documented; these include crossbite, crowding, retrusion, palatal arch contracture, lingually inclined mandibular teeth, and mandibular arch contracture.

A prosthodontist should fabricate and deliver commissure splints within days of any trauma to prevent microstomia, considering that wound margins begin to contract 5 days after injury. The prosthesis consists of an acrylic stent with acrylic conformers curving laterally and posteriorly to retract the corners of the mouth to simulate the desired contours of the commissures. The prosthesis is worn 24 hours a day for 6 to 8 months with an additional 4 to 6 months of nighttime wear.²⁵

Auricular Prostheses

Extraoral prostheses may be the treatment of choice for patients who do not want to undergo surgery, patients with defects that cannot be predictably reconstructed surgically, or as an interim prosthesis if surgical reconstruction must be delayed. The prosthetic rehabilitation of auricular defects, both congenital and acquired, requires consultation with a plastic surgeon to determine whether an acceptable surgical result can be achieved. Silicone elastomers and other synthetic materials have made it possible to fabricate natural-appearing prostheses. Temporary prostheses made of heat-polymerizing acrylic may be fabricated and delivered to the tissue bed 3 to 4 weeks after surgery. Acrylic resin prostheses allow periodic adjustment and relining with denture soft reline material to accommodate changes in the soft tissue.

If reconstruction is not anticipated, the auricle and any remaining tissue tags should be removed to improve retention of the prosthesis, positioning and contour relative to the tragus, and ease of placement by the patient. The tragus should be retained or surgically reconstructed, if necessary, to conceal the anterior tissue margin of the prosthesis. After complete resection of the auricle, a nonmobile, flat tissue bed lined with split-thickness skin, full-thickness skin, or a pedicle flap provides the most favorable base for an auricular prosthesis. Flaps containing hair should be avoided, because most auricular prostheses are retained with adhesives, and hair complicates cleaning implant components in implant-supported prostheses and interferes with margin integrity.

Defects resulting from the partial resection of the auricle or microtia are more difficult to restore prosthetically. The margins of a prosthesis tend to be more visible, and patients may experience greater difficulty orienting a prosthesis over a partially retained ear, which may be malformed.

Medical grade adhesives are often used to retain auricular prostheses; however, some patients may encounter problems, such as retention, skin irritation, and prosthesis misalignment. The use of bone-anchored osteointegrated implants to retain an extraoral prosthesis can eliminate retention problems and provide aesthetic restoration.

Trismus Rehabilitation

Patients must consult with a reconstructive surgeon before they begin a postoperative exercise program for the mandible. Patients should perform mandibular exercises multiple times each day, and progress should be documented by the patient, physical therapist, and prosthodontist. Maximal opening of the mandible with movement away from the operated side loosens scar tissue, reduces trismus, and improves the patient's ability to achieve a more normal maxillomandibular relationship. Devices are available to counteract limited jaw opening during the immediate post-surgical period, minimize contracture of scar tissue, strengthen jaw muscles, and mobilize the joints. A simple and inexpensive technique is to use tongue blades to gauge the degree of mouth opening between the anterior teeth and repeat this several times during the day. A threaded, acrylic, tapered screw can also be used, which can be alternately placed on the anterior or posterior teeth and gradually turned to increase opening of the jaw. Commercial devices, such as the TheraBite Jaw Motion Rehabilitation System (Atos Medical) or the Dynasplint System (Dynasplint Systems), are available to mechanically assist restoration of proper jaw opening. These devices allow a patient to comfortably control the degree of opening with repetitive passive motion, stretch the jaw in an anatomically correct pattern, and measure the degree of opening.

CONCLUSION

The application of principles in orthodontics and maxillofacial prosthetics has many indications in pediatric maxillofacial surgery. It is our hope that this chapter informs the reader of the available resources of his or her dental colleagues. The growing pediatric patient with a mixed dentition may not be a candidate for a definitive surgical solution. Frequently maxillofacial prosthetics provide an excellent alternative to a surgical procedure that creates both an aesthetic and functional result. Additionally, the specialty of orthodontics continues to improve the ability to modify the facial skeleton orthopedically using advanced designs in headgear and skeletal anchorage. Knowledge and application of these advances and principles to the pediatric population will help ensure that each patient achieves the best result for their clinical problem.

KEY POINTS

- Many oral problems seen in the pediatric population can be treated nonsurgically, and treatments may be enhanced when appropriate dental specialists are involved in a patient's care.
- For patients requiring surgery and prosthetic restoration, the plastic surgeon should refer the patient for prosthetic evaluation preoperatively to determine the type and design of the restoration based on the configuration of the defect.

- Angle's concept of normal occlusion remains effective today and is the primary occlusal relationship desired in most modern orthodontic cases. It is evident that if molar relationships are ideal, and the teeth are of proportionate size, the remaining dentition will align in an ideal relationship. Although there is some debate regarding the definition of "ideal" occlusion, most investigators find that this relationship is the most aesthetic as well as "protective," because it minimizes aberrant forces.
- A diagnosis of complex maxillofacial problems involving occlusion is based on history, physical examination, radiographic examination, and dental casts accurately mounted on an articulator.
- Cephalometric radiographs are useful to evaluate the relationship of teeth to underlying bone, the upper and lower jaws to one another, and jaws to the cranial base. Careful review of cephalometric radiographs in the context of the patient's history and physical examination findings is very useful to elucidate the true nature of the problem. It also serves as a useful tool for planning the surgical reconstruction.
- Panoramic radiographs provide useful information, such as interproximal bone height, quality of the bone in the maxilla and mandible, locations of plates and screws, sinus configuration, location of the inferior alveolar canal, anatomy of the mandibular condyles and eminences, impacted teeth, missing teeth, and significant areas of dental decay and periapical disease.
- The DentaScan images the jaws in thin slices in the coronal plane (viewed from third molar to third molar), allowing one to view each tooth and the surrounding bone in millimeter increments.
- The role of presurgical orthodontics varies from early alignment of dentition, establishing proper incisor positions, and creating a compatible interarch relationship, to later preparation of the arch before placement of osteointegrated implants.
- Before deciding which prosthesis should be used, the surgeon must consider the patient's age, skeletal growth, occlusion, periodontal status, tooth eruption patterns, orthodontic treatment, quality and quantity of hard and soft tissue, oral hygiene, psychological state, and preference for a removable or fixed prosthesis.
- Osteointegrated implants are often the treatment of choice for skeletally mature patients when anterior teeth neighboring the edentulous space have a history of minimal or no restorative treatment. A removable appliance is recommended for pediatric patients who have tooth loss, because the prosthesis can be adapted and adjusted with ease according to growth, dental changes, and bone grafting procedures to the edentulous area.

Continued

KEY POINTS (continued)

- Obturator prostheses are indicated for patients who have congenital or acquired tissue openings, primarily of the hard palate and/or contiguous alveolar structures. Obturator prostheses may require an additional speech aid component to manage velopharyngeal insufficiency caused by defects of the hard and soft palate.
- A speech aid prosthesis (soft palate obturator) is a maxillary removable prosthesis indicated for restoring acquired or congenital/developmental aberrations of the soft palate, with a portion extending into the pharynx to separate the oropharynx and nasopharynx during phonation and deglutition, thereby completing the palatopharyngeal sphincter. Speech aid prostheses are indicated for selected patients diagnosed with velopharyngeal insufficiency.
- A palatal lift prosthesis differs from a speech aid prosthesis in that it does not extend into the pharynx to restore palatopharyngeal function. The palatal lift elevates the soft palate (adequate length and size) superiorly and posteriorly to achieve soft palate closure with the peripheral pharyngeal tissues, thereby improving voice quality by increasing oral pressure for consonant articulation and reducing hypernasality and escape of nasal air.
- Extraoral devices such as auricular prostheses may be the treatment of choice for patients who do not want to undergo surgery, for defects that cannot be predictably reconstructed surgically, or if an interim prosthesis is desired before a delayed surgical reconstruction.

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30

Craniofacial Microsomia

Lee W.T. Alkureishi • Rosemary Seelaus • Pravin K. Patel



Three-Dimensional Imaging for Craniofacial Microsomia Patients; Virtual Surgical Planning: Asymmetrical LeFort I Advancement and Bilateral Sagittal Split Osteotomy.

THE NOMENCLATURE OF CRANIOFACIAL MICROSOMIA

Craniofacial microsomia (CFM) comprises the spectrum of craniofacial anomalies that results in underdevelopment of the facial structures affecting the hemiface, either unilateral or bilateral, to a varying extent. These conditions are known by a variety of names because of the wide range of phenotypic expression (Fig. 30-1 and Box 30-1). The names include *first and second branchial arch syndrome*, *lateral facial dysplasia*, *otomandibular dysostosis*, *oculoauriculovertebral dysplasia* (*Goldenhar syndrome*), and within the Tessier clefting classification, a *number 7 cleft*. Patients with unilateral microtia and mandibular deficiency typically are described as having hemifacial microsomia (HFM). However, those who also have epibulbar dermoids and hemivertebral anomalies have Goldenhar syndrome.^{1,2} Some patients have only isolated components within the spectrum, such as preauricular tags and microtia without other obvious anomalies. Similarly, patients may present with lateral facial clefting (macrostomia) without other obvious anomalies or with hemimandible hypoplasia without overlying soft tissue involvement.



Fig. 30-1 The variability in the phenotypic expression of CFM. **A**, Right mandibular hypoplasia (Kaban-Pruzansky type I) (see Table 30-3). **B**, Right mandibular and maxillary deformity and right microtia (Kaban-Pruzansky type IIA). **C**, Right mandibular and maxillary deficiency, right microtia, right marginal nerve palsy, and cervical scoliosis (Kaban-Pruzansky type IIB). **D**, Right mandibular and maxillary deficiency, right orbital dystopia, right microtia, and right macrostomia (Kaban-Pruzansky type III). These patients will appear later in the chapter.

Box 30-1 Terms Used Synonymously With Craniofacial Microsomia

- Hemifacial microsomia
- Goldenhar syndrome
- Oculoauriculovertebral spectrum
- Otomandibular dysostosis
- First and second branchial arch syndrome
- Auriculobranchiogenic dysplasia
- Facioauriculovertebral dysplasia
- Unilateral mandibulofacial dysplasia
- Facioauriculovertebral malformation complex

In most patients, the condition is unilateral; thus the term *hemifacial microsomia* is most commonly used. However, 15% to 30% of cases have bilateral involvement (bilateral HFM) and usually have differential involvement with asymmetrical hypoplasia.² Therefore the term *craniofacial microsomia* is more appropriate, and the two subtypes are distinguished as unilateral CFM and bilateral CFM. Although CFM is a more inclusive description and encompasses the wider spectrum of presentation, it fails to fully address the small subset of patients with associated extracranial anomalies. Clinicians and researchers are required to supplement the simplistic label with a fuller description that includes the affected components. An increasingly comprehensive classification schema has evolved.

EPIDEMIOLOGY

CFM is the second most common group of birth defects after cleft lip and palate, with an incidence of approximately 1 in 5500 live births. This incidence may be underestimated, however, with many of the milder forms of the condition potentially unrecognized.^{3,4} Some authors have suggested that isolated microtia should be considered a microform of HFM, which may further affect current incidence estimates.⁵

In a multicenter retrospective study, the odds ratio for developing CFM was 9 for multiple gestations (19 if infertility treatment was used), 6 if birth weight was less than 2000 g, and 5 if the family has a history of CFM, auricular abnormalities, or hearing loss.⁶ CFM was 40% more common in males and occurred more often on the right side. Low family income, low maternal weight (less than 18 kg/m²), discovery of pregnancy after 9 weeks, and previous termination or spontaneous abortion were each estimated to independently increase the risk of CFM. The risk of CFM was significantly lower for black mothers and was borderline increased for Native American mothers.

In a follow-up study, Werler et al⁷ examined first trimester exposure to vasoactive medications, including pseudoephedrine, phenylpropanolamine, aspirin, and ibuprofen. The authors found increased odds ratios associated with pseudoephedrine exposure, alcohol use, and the combined use of vasoactive medications and tobacco.

PATHOGENESIS

The underlying mechanism of CFM development is not fully understood but is thought to result from abnormal development of the first and second branchial arches. In normal development, the first branchial arch forms Meckel cartilage, which further develops into the mandible, muscles of mastication, most of the malleus, and the incus. The second branchial arch forms Reichert cartilage, which develops into the hyoid bone, styloid process, muscles of facial expression, and most of the stapes (Fig. 30-2).

In the most widely accepted theory of pathogenesis, a vascular insult impairs development of these first and second arch derivatives. Poswillo⁸ demonstrated features consistent with CFM in mice and monkeys by inducing hematomas in the region of the stapedia artery. The resultant tissue necrosis and variable regeneration are thought to explain the broad spectrum of features observed.

Retinoic acids produce anomalies in the axial skeleton, the craniofacial region, the cardiovascular system, and the central nervous system, similar to those seen in HFM.⁹

The discovery of a twin with microtia whose co-twin was resorbed in utero supports the hypothesis that some cases of HFM may be caused by the transfer of thromboplastin-rich blood from a dead to a living monozygotic or dizygotic twin through placental vascular channels.¹⁰

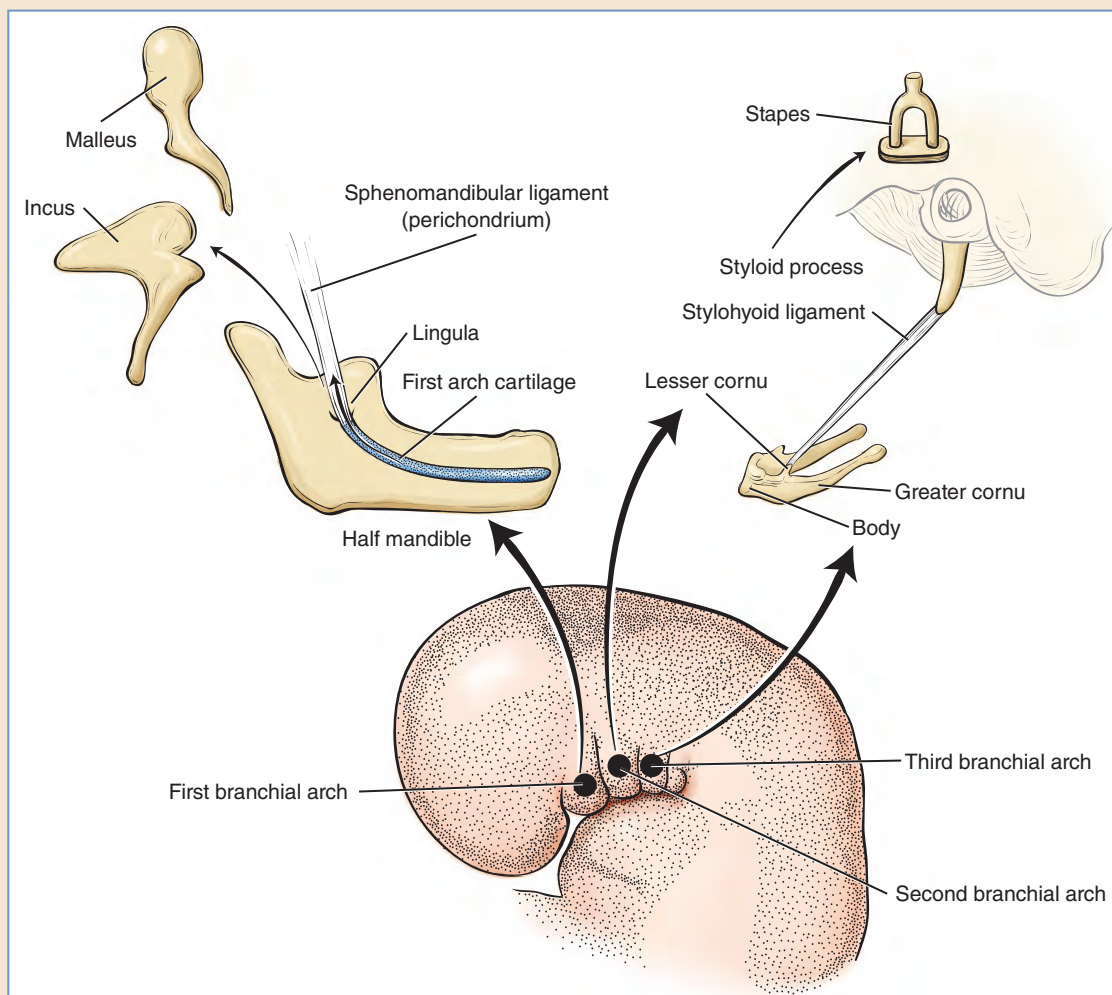


Fig. 30-2 Derivatives of the first, second, and third branchial arch cartilages.

GENETIC ASPECTS

Growing evidence supports a genetic basis for at least a proportion of HFM cases. Although most occurrences appear to be sporadic, a subgroup of patients have a family history of similar phenotypic characteristics. This subgroup was previously thought to be approximately 2% of HFM cases; however, more recent studies suggest that the incidence may be as high as 45%. This discrepancy may result from epigenetic mechanisms and/or variable expression, leading to under-recognition of mild phenotypic features in previous studies.^{4,11}

Studies examining familial HFM have shown a variety of inheritance patterns, including autosomal dominant and autosomal recessive.^{12,13} A variety of discrete chromosomal anomalies have been identified in patients with phenotypes consistent with the HFM spectrum. A chromosome 5p15 deletion, 12p13 deletion (*WNT5B* gene), partially overlapping microduplications on

14q23.1, anomalies on chromosomes 22q and 14q32 (*GOOSECOID* gene), and others have been implicated in the development of HFM.¹⁴⁻¹⁶

Despite these findings, the various gene defects have not yet been linked by a common molecular mechanism. The pathogenesis of HFM and its variants is probably as diverse as its phenotypic expression. Although Mendelian inheritance is possible in familial cases, sporadic cases may have multifactorial causes. This was shown by multiple studies demonstrating an increased incidence of HFM associated with maternal diabetes, smoking, twin pregnancies, and certain medications such as thalidomide and phenylephrine.¹⁶

Genetic counseling for families of children with HFM should include a discussion of the risk of sibling recurrence. In the absence of a family history and with a normal karyotype, this is approximately 2% to 3%.¹⁶ A thorough family history-taking is critical, because minor manifestations of the condition in relatives may have gone unrecognized and can potentially increase the risk of sibling recurrence.

THE CLINICAL SPECTRUM

Clinical features of CFM affect the various skeletal, muscular, neural, and soft tissue components to a varying extent¹⁷ (Table 30-1). A thorough physical examination with objective studies is essential given the wide spectrum of anatomic and corresponding functional abnormalities. A comprehensive evaluation involves multiple specialties, including genetics, plastic and craniofacial surgery, otolaryngology, audiology, anaplastology, speech and language pathology, psychology, ophthalmology, orthopedics, and dentistry (orthodontics and prosthodontics).

Table 30-1 Principle Craniofacial Anomalies of Craniofacial Microsomia

Facial Region	Abnormality	Incidence (%)
Mandible	Mandibular hypoplasia	89 to 100
	TMJ maldevelopment	24 to 27
Ear	Microtia	66 to 99
	Preauricular remnants	34 to 61
	Conductive hearing loss	50 to 66
Midfacial	Maxillary hypoplasia	
	Zygomatic hypoplasia	
Dental	Maldevelopment	
	Dental crowding	
Eye	Orbital dystopia	
	Microphthalmia	
	Epibulbar dermoids	
Mouth	Macrostomia	17 to 62
Muscle	Muscle hypoplasia	85 to 95
	Levator muscle hypoplasia	
Nerve	Cranial nerve VII palsy	
Soft tissue	Subcutaneous hypoplasia	

TMJ, Temporomandibular joint.

Skeletal Involvement

Although the extent of the deformity may include the entire hemicraniofacial skeleton, the mandibular deformity is considered the cornerstone (Fig. 30-3), and asymmetrical mandibular growth and development is the presenting complaint in most of patients. The posterior body of the mandible, the ascending ramus, the coronoid, and the condylar process have varying degrees of hypoplasia. The most severe cases have complete absence of the ramus and correspondingly the temporomandibular joint (condyle and glenoid complex). In such cases, the zygomatic arch may be absent and the temporal base hypoplastic.

The mandible plays a pivotal role in progressive distortion of the facial skeleton on the affected side, and secondary compensation occurs on the contralateral side. The natural history is progressive deviation toward the affected side, until the end-stage deformity is established at skeletal maturity. The normal downward vertical growth of the maxilla is restricted by the hypoplas-

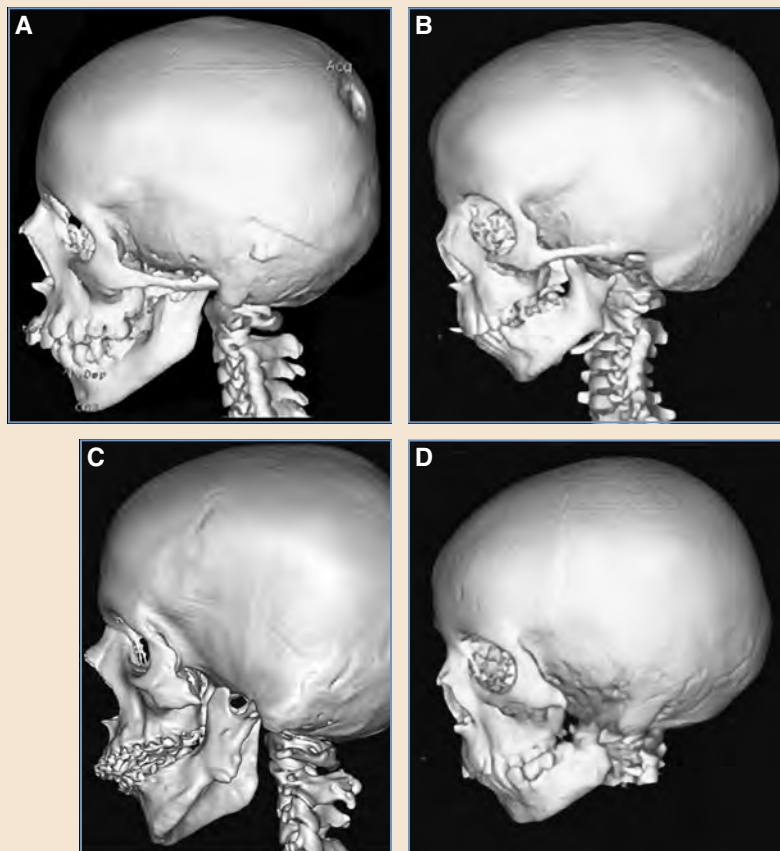


Fig. 30-3 The variability in the skeletal deformity of CLP is shown in a series of CT scans. Common to all of the cases is a mandibular deformity with various components affected. **A**, Kaban-Pruzansky type I. **B**, Kaban-Pruzansky type IIA. **C**, Kaban-Pruzansky type IIB. **D**, Kaban-Pruzansky type III.

tic mandible, resulting in multiplanar deficiency of the hemimaxilla and obliquity of the occlusal plane (coronal roll, transverse yaw, and sagittal pitch). The zygomatic arch, with the glenoid fossa, may range from fully present to rudimentary to completely absent. The zygomaticoorbital complex is affected in up to 15% of patients, with varying degrees of inferior, medial, and rotational displacement of the orbit. Microphthalmia may be present. In more extensive cases, the involvement extends to the cranial vault (frontal bone) and skull base (temporal bone). Malformations of the cervical vertebrae can occur and include the presence of hemivertebrae and fused vertebrae.

In the cases of CFM with involvement of the orbit, cranium, and vertebrae (Fig. 30-4), clinicians should not focus solely on the mandibular asymmetry. Interventions should be performed within the context of the entire spectrum of involvement. Correction of the mandibular-maxillary relationship does not necessarily correct the upper midfacial involvement, and symmetry with respect to the absolute reference plane of the skull base must be clearly understood.

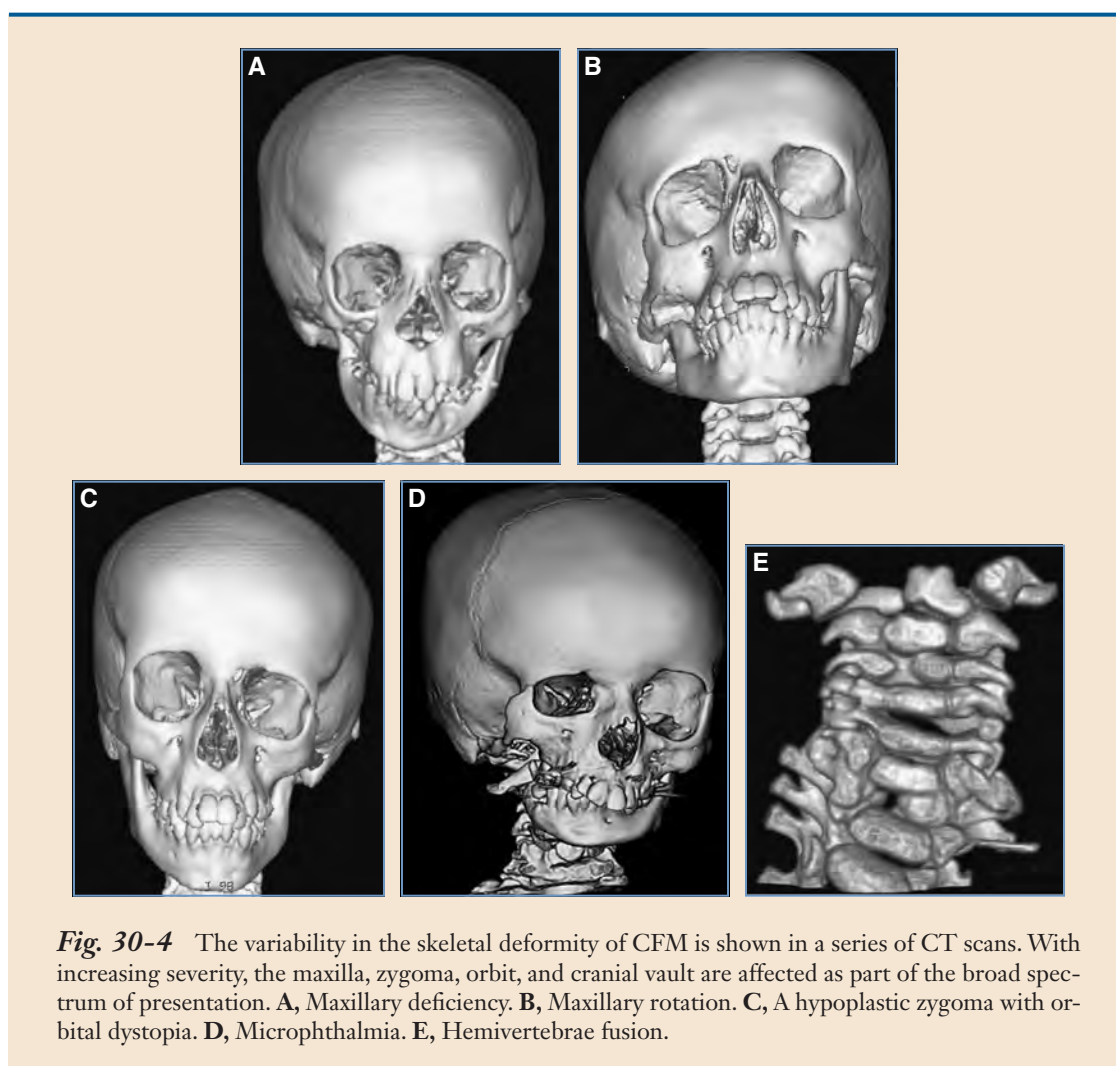


Fig. 30-4 The variability in the skeletal deformity of CFM is shown in a series of CT scans. With increasing severity, the maxilla, zygoma, orbit, and cranial vault are affected as part of the broad spectrum of presentation. **A**, Maxillary deficiency. **B**, Maxillary rotation. **C**, A hypoplastic zygoma with orbital dystopia. **D**, Microphthalmia. **E**, Hemivertebrae fusion.

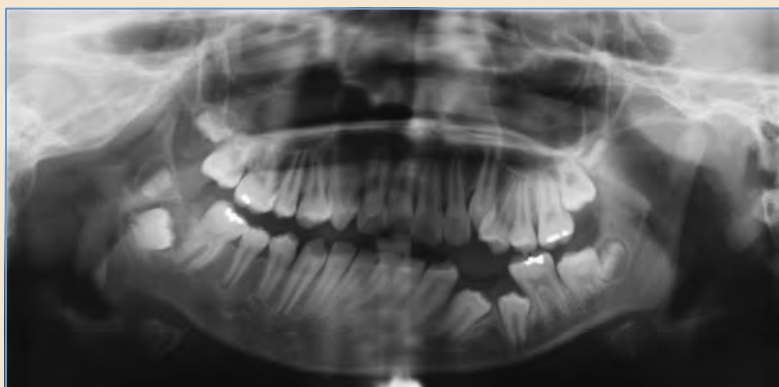


Fig. 30-5 An ortho-Panorex radiograph shows the dental crowding and crossbite that accompany a hypoplastic mandible.

Dental Involvement

Skeletal hypoplasia, primarily of the mandible, results in dental crowding, crossbite, and deviation of the anterior teeth toward the hypoplastic side.¹⁸ Underdevelopment of the posterior mandibular body region correspondingly affects the posterior molar development, and in severe cases, the posterior dentition will be absent and require prosthodontic replacement (Fig. 30-5). In some cases the condition may also be associated with enamel defects and hypodontia, and patients may be at increased risk for early childhood caries.¹⁹

Muscular Involvement

The temporalis muscle normally inserts into the coronoid process, the lateral pterygoid muscle inserts into the condyle, and the medial pterygoid inserts into the mandibular angle. However, when the skeletal components of the mandible are diminutive or absent, then the corresponding muscle origins and insertions are affected (Fig. 30-6). When the zygomatic arch is absent, the origin of the masseter is affected, and in such cases the temporalis and masseter muscle may become confluent. Functionally, the chin deviates to the affected side with opening and closing. Thus symmetry achieved with skeletal reconstruction, including construction of a pseudotemporomandibular joint, is only a static symmetry and cannot restore normal symmetrical oromotor function. Lateral gnathism always occurs, except in the mildest forms of CFM. The muscular involvement extends intraorally, and in some cases, muscular hypoplasia of the levator palatini on the affected side and asymmetrical functioning result in velopharyngeal insufficiency.²⁰

Neural Involvement

The branches of the facial nerve can be involved to varying degrees. The marginal mandibular nerve is most often involved, resulting in lower lip asymmetry that is distinctly more noticeable with smiling (Fig. 30-7). The next most commonly involved branches are the temporal and zygomatic branches, affecting eyelid closure and placing patients at risk for exposure keratitis.

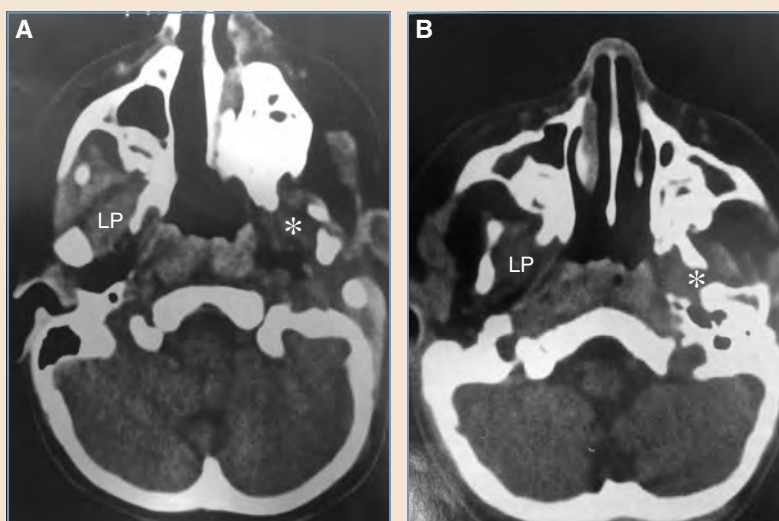


Fig. 30-6 A cross-sectional CT scan reveals the hypoplastic muscle that accompanies the skeletal deformity as it affects the normal origin and insertion. Absence of the zygomatic arch would affect the masseter muscle. Absence of the coronoid would affect the temporalis muscle. Absence of a condyle would affect the lateral pterygoid muscle, as shown in **A**, Pruzansky grade II and in **B**, Pruzansky grade III deformities. Absence of the coronoid would affect the temporalis muscle. (*, Affected side; LP, lateral pterygoid muscle.)



Fig. 30-7 This patient has involvement of the marginal mandibular nerve on the left side with resulting asymmetrical lower lip muscular function.

Auricular Involvement

Auricular involvement ranges from the presence of preauricular tags to complete absence of the external ear (anotia) (Fig. 30-8). Within these extremes, microtia is seen in 66% to 99% of patients with CFM and is usually associated with a 50- to 55-decibel hearing loss on the affected side.^{17,21} Atresia of the external auditory canal and middle ear abnormalities are frequent findings and are associated with varying degrees of conductive hearing loss. Less often, abnormalities of the in-

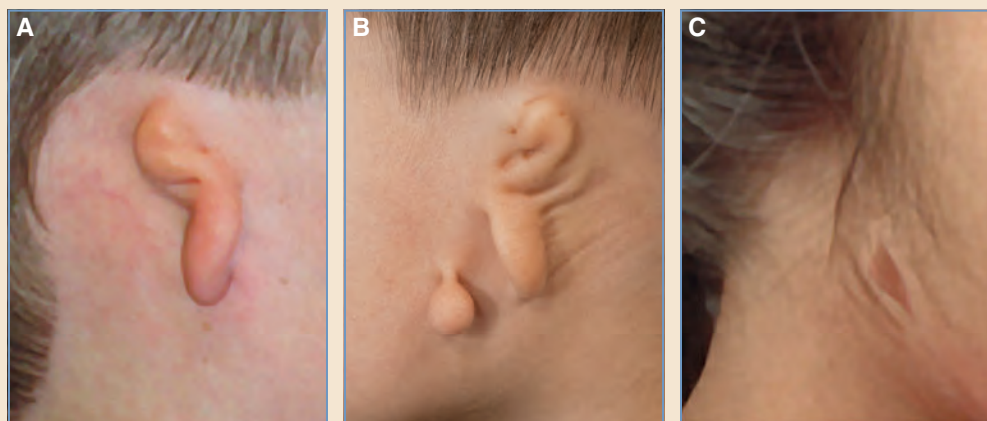


Fig. 30-8 CFM has a broad range of auricular involvement, from anotia to an unaffected ear. **A** and **B**, Lobular-type microtia. **C**, Anotia.

ner ear canal, cochlea, and semicircular canals occur and may lead to sensorineural hearing loss and/or balance disturbances.^{22,23} These malformations of inner ear structures are not explained by a defect of the first and second branchial arches, and additional pathogenic mechanisms such as failure of neural crest migration and a defect of blastogenesis have been proposed as contributing factors.^{24,25} The severity of external ear deformity has been shown to correlate closely with the degree of middle ear malformation, whereas temporal bone abnormalities are predicted by the severity of mandibular hypoplasia.^{26,27} Temporal bone CT is helpful in delineating the dysmorphology of the middle and inner ear structures, and grading the extent of abnormal anatomy (Jahrsdoerfer classification) can help to guide surgical planning.²⁸ In patients with unilateral CFM, hearing in the normal-appearing, contralateral ear is usually unaffected. However, a few patients have an associated conductive or mixed hearing loss on the contralateral side, thought to result from the presence of effusions and otitis media.²⁹ Identifying patients with bilateral hearing loss early is crucial, because early intervention is required to prevent problems with speech and language development. Every patient with atresia of the external ear canal should therefore undergo an early audiologic evaluation to determine the type and severity of hearing loss.

Ocular Involvement

Ocular findings are present in approximately two thirds of patients with CFM and are more common in patients with the Goldenhar syndrome variant³⁰ (Fig. 30-9). Epibulbar dermoids, a form of choristoma, are the most frequent finding and are usually seen in the inferotemporal location. Conjunctival lipodermoids (dermolipomata) are more yellow in color compared with epibulbar dermoids and arise superotemporally or inferiorly.³¹ Upper eyelid colobomas are seen in up to 24% of Goldenhar patients, most frequently at the junction of the middle and central thirds, and are usually quadrangular in shape.³² Colobomas require urgent attention, because failure to protect the globe can lead to corneal abrasion and subsequent scarring.

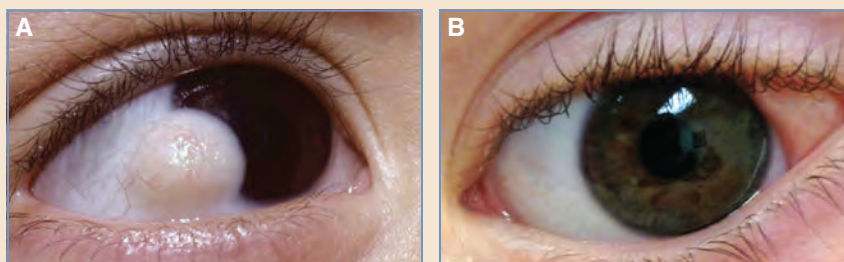


Fig. 30-9 Ocular findings in patients with CFM. **A**, An epibulbar dermoid. **B**, A lipodermoid.



Fig. 30-10 This patient has macrostomia (a Tessier number 7 cleft) as a component of CFM.

The function of the levator palpebrae muscle may be compromised in these patients, leading to congenital ptosis in 12% of patients with Goldenhar syndrome.³³ Additional ocular findings can include blepharophimosis, atresia of the nasolacrimal drainage system, strabismus, microphthalmia, and anophthalmia. The caruncles may be dysplastic, and colobomas can occur on the iris, optic nerve, and within the chorion or retina. These ocular features are usually unilateral but are bilateral in a small percentage of cases.³² An associated facial nerve palsy may lead to lagophthalmos, with the attendant risks of corneal drying and ulceration.

Lip Involvement

A lateral or transverse facial cleft (Tessier number 7) is present in approximately one quarter of patients with CFM and manifests as macrostomia (Fig. 30-10). This finding is five to six times more likely to be unilateral and is usually on the same side of the face as the other predominant features of CFM.³⁴ The severity ranges from mild widening of the oral aperture with loss of commissural anatomy to a full-thickness cleft running from the mouth to the tragus. The cleft may affect skin, subcutaneous fat, muscles of mastication, facial expression, and buccal continence. Severe cases

Table 30-2 Classification of Macrostomia

Type	Features*
I	Minor unilateral macrostomia
II	Major unilateral macrostomia
III	Minor bilateral macrostomia
IV	Major bilateral macrostomia

*The distinction between minor and major macrostomia is made when the cleft extends laterally to the anterior border of the masseter muscle.

can have underlying bony discontinuity. Labial clefting is also associated with greater severity of orbital involvement.³⁴ In a series of 12 patients with macrostomia, Gleizal et al³⁵ reported that the extent of muscular clefting can continue sagittally to include the tonsillar pillar or laterally to involve the masseter, and any repair must address these deficiencies. The authors proposed a surgical classification scheme that includes these features³⁵ (Table 30-2).

Cleft lip and palate deformity is reported in 10% of patients with CFM and, similar to patients with a Tessier number 7 cleft, occurs on the predominantly affected hemiface. The management of this deformity in patients with CFM is similar to that for patients with isolated cleft lip and palate and is described in Chapter 19. The presence of labial clefting, either in the form of a cleft lip or a lateral facial cleft, can lead to oral incompetence with attendant feeding difficulties, salivary incompetence, and later, speech problems, and warrants early correction.

Soft Tissue Involvement

Although much of the deformity involves the skeletal framework, skin and subcutaneous tissue are also involved with varying degrees of volumetric and corresponding skin surface area deficiency. The parotid gland is deficient in some patients. Restoring symmetry requires addressing the tissue “upholstery” of the skeletal framework.

Extracranial Involvement

Associated extracraniofacial anomalies can involve multiple organ systems, including the central nervous system, heart, skeleton, lungs, gastrointestinal tract, and kidneys. These are listed in Box 30-2.³⁶

OBJECTIVE STUDIES

Abnormal growth and development of the craniofacial structures and the impact of intervention must be properly documented to define the anatomic extent of the deformity and its functional effect. This is essential to optimize treatment planning and assess outcomes.

Standardized, two-dimensional digital photographs are needed to document not only the extent, but also the progression of facial deformity from infancy to adulthood. The recently published Phenotypic Assessment Tool for CFM provides an example of a standardized photographic

Box 30-2 Extracranial Anomalies in Craniofacial Microsomia**Skeletal**

Scoliosis
 Hemivertebrae/vertebral anomaly
 Rib anomaly
 Congenital hip deformity
 Pectus excavatum
 Chest wall deformity
 Hyposegmental sternum
 Short, webbed neck
 Anomalous limb
 Absent or anomalous thumb
 Radial/ulnar hypoplasia
 Toe syndactyly
 Clubfoot
 Digital hypoplasia
 Digital polydactyly
 Digital syndactyly
 Pes cavum
 Valgus deformity
 Leg rotation

Cardiac

Murmur
 Ventricular septal defect
 Atrial septal defect
 Anomalous pulmonary return
 Tetralogy of Fallot
 Patent ductus arteriosus
 Pulmonary stenosis
 Aortic stenosis
 Transposition of great vessels
 Vascular ring
 Acyanotic “heart” disease
 Dextrocardia

Central Nervous System

Hydrocephaly
 Hemiparesis
 Microcephaly/partial anencephaly
 Agenesis of corpus callosum
 Arnold-Chiari malformation
 Dandy-Walker syndrome/encephalocele
 Seizure disorder

Pulmonary

Tracheoesophageal fistula
 Hypoplasia
 Choanal atresia
 Stenotic trachea

Gastrointestinal

Bilateral inguinal hernia
 Gastroesophageal reflux
 Displaced or absent rectum
 Imperforate anus
 Intestinal malrotation

Renal

Size discrepancy
 Agenesis
 Ectopic
 Reflux
 Rotated
 Conjoined

protocol³⁷ (Fig. 30-11). Although two-dimensional photographs are the minimum standard, the complex asymmetry of patients with CFM is best captured, documented, and analyzed with three-dimensional image-capture devices and photogrammetry (Fig. 30-12). When possible, such imaging should be performed annually and more frequently when rapid changes are occurring in early childhood development and with surgical intervention. Currently available CT scans with three-dimensional reconstruction are indispensable to surgeons for fully detailing the skeletal involvement, for defining the complex asymmetry in all three planes, and for surgical planning (Fig. 30-13). Moreover, they are the only objective tool for measuring the outcomes of surgical intervention.³⁸



Fig. 30-11 A standardized, two-dimensional photographic record as part of the Phenotypic Assessment Tool for patients with CFM.



Fig. 30-12 Images of a patient with CFM captured using a three-dimensional camera system (see the accompanying video). The digital data provide linear surface distance, surface areas, and volumes that can be measured.

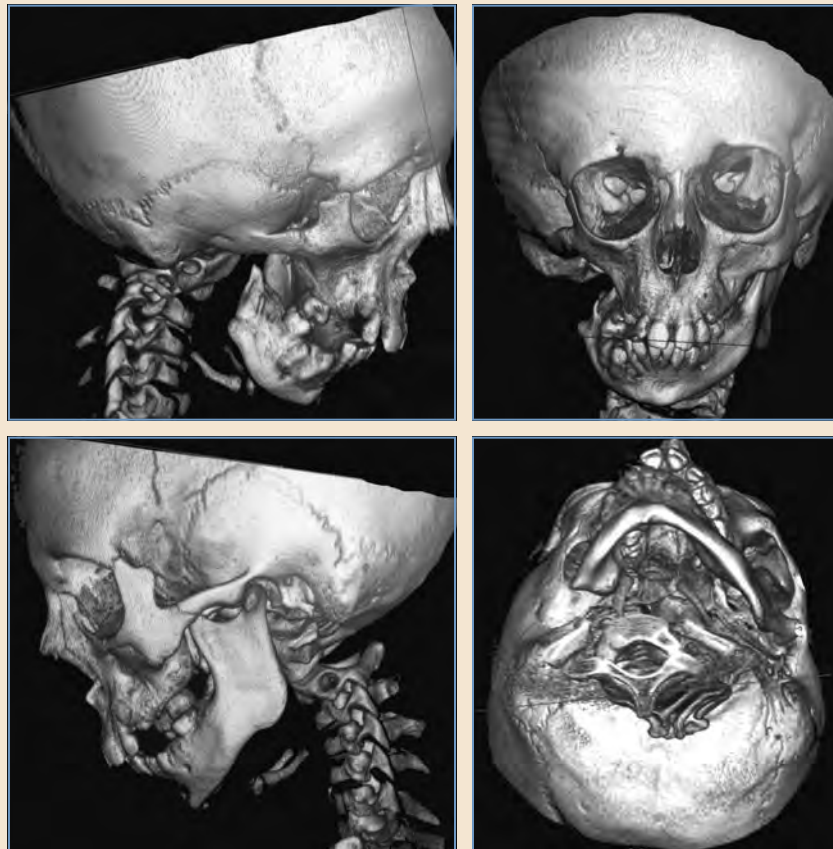


Fig. 30-13 A three-dimensional craniofacial CT scan of a patient with CFM and asymmetry in all three planes.

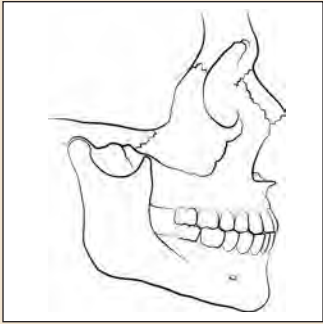
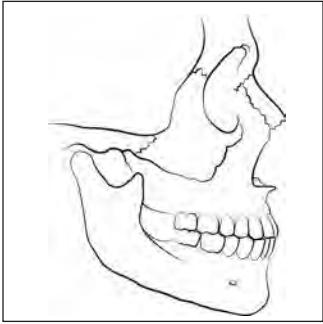
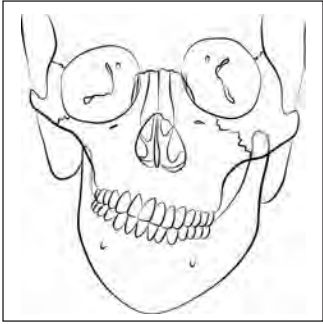
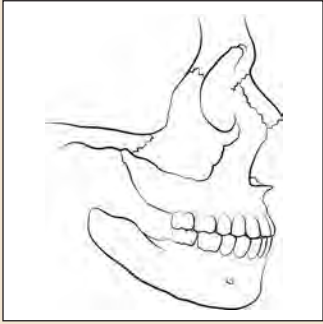
CFM can be diagnosed based on clinical examination; however, the impact of the embryologic fault on the underlying structures cannot be fully assessed. In a recent review of 41 patients, Wink et al³⁹ demonstrated poor correlation between clinical scoring according to the Kaban-Pruzansky classification (see Table 30-3) and classification based on three-dimensional CT imaging. They questioned the reproducibility of the currently accepted method. We prefer to obtain a three-dimensional CT scan at the time of the initial evaluation to fully establish the extent of involvement and to guide overall treatment planning. At subsequent time points, we prefer to use the lower radiation dose of a cone-beam CT scan primarily for orthodontic intervention and a conventional, medical three-dimensional CT scan when high-resolution detail is needed that may affect surgical intervention. In most instances we can limit the radiation dosage without ultimately affecting outcome.

A number of functional studies are relevant to children with CFM. Annual audiograms are essential to maintain and monitor the hearing of the unaffected side. Testing is needed to assess the value of bone conduction hearing devices, which would be beneficial in many children with CFM to provide binaural hearing. Nasendoscopy is essential for assessing velopharyngeal function, and nasorhinometry is beneficial for evaluating the upper airway. Many children may have obstructive sleep apnea, and this is best documented by a formal sleep study. Neurodevelopmental and quality-of-life assessment tools, administered by psychologists, help to assess well-being.

CLASSIFICATION

The broad spectrum of phenotypic variability has led to a number of classification systems for CFM. Of the various skeletal classification schemes, the Pruzansky classification⁴⁰ of mandibular involvement, described in 1969, and its subsequent modification by Kaban et al⁴¹ in 1988 (Table 30-3) provide the most workable schemas for treatment planning. In Pruzansky grade I, all of the anatomic elements of the mandible are present, but they are of diminutive volume, compared with the unaffected side. In Pruzansky grade III, the ascending ramus, the coronoid process, the condyle, and the corresponding glenoid fossa are absent. The mandible tapers at what would be the angle in a normal mandible. Between these two extremes, all remaining hypoplastic mandibles (ramus, condyle, and coronoid process) are classified as Pruzansky grade II. The later modification by Kaban et al⁴¹ further subdivided Pruzansky grade II into type IIA, in which the glenoid fossa–condyle relationship is spatially maintained at the cranial base, and type IIB, in which the mandible and the corresponding glenoid fossa, if present, are displaced medially.

Table 30-3 Mandibular Classifications

Pruzansky Classification	Description	Illustration	Kaban-Pruzansky Classification
Grade I	Ramus, condyle, and glenoid are all distinctly present, but smaller compared to the unaffected side.		Type I
Grade II	Ramus, condyle, and coronoid are present each to a varying extent but are hypoplastic and abnormally shaped.		Type IIA
	Ramus, condyle, and coronoid are present each to a varying extent but are hypoplastic and abnormally shaped.		Type IIB
Grade III	Complete absence of ramus, glenoid fossa, and TMJ. The mandibular body length is also typically short on the affected side.		Type III

The Pruzansky classification⁴⁰ and the modification by Kaban et al,⁴¹ which describes two subtypes of Pruzansky grade II based on the temporomandibular joint morphology (*TMJ*) and position. These help to direct the surgical treatment and affect the outcome. Type IIA has a well-defined articulation that acts as a superior “stop” for mandibular distraction and conventional orthognathic surgery.

Table 30-4 Classification of Hemifacial Microsomia

Type	Description
Type IA	The craniofacial skeleton is only mildly hypoplastic, and the occlusal plane is normal.
Type IB	The craniofacial skeleton is as in type IA, but the occlusal plane is canted.
Type II	The condyle and part of the affected ramus are absent.
Type III	In addition to the findings in type II, the zygomatic arch and glenoid fossa are absent.
Type IV	This type is uncommon, with hypoplasia of the zygoma and medioposterior displacement of the lateral orbital wall.
Type V	This is the most extreme type, with inferior displacement of the orbit and a decrease in orbital volume.

The classification, although limited to the mandible, is helpful for managing lower skeletal asymmetry. Lauritzen et al⁴² proposed a schema that addressed the midfacial and orbital skeletal asymmetry that accompanies the mandibular deficiency (Table 30-4).

More inclusive systems include the Skeletal, Auricular, and (Soft) Tissue classification, and the Orbit, Mandible, Ear, Nerve, and Soft Tissue (OMENS) classification.^{3,43} The OMENS classification is the most widely used in practice and has undergone refinement to include extracranial anomalies (now called *OMENS-Plus*)⁴³ (Table 30-5) and pictorial representations of the various deformities^{36,37,44} (Fig. 30-14). Using the OMENS-Plus system, each extracranial anomaly is recorded by organ system and counted once; for example, OMENS + 2 cardiac (1 = atrial septal defect [ASD]; 2 = ventricular septal defect [VSD]). Gougoutas et al⁴⁴ presented a more recent pictorial classification of OMENS that provides an excellent visual tool. Compared with the more simplistic Pruzansky classification, the more comprehensive classifications are not as clinically practical in guiding the surgical treatment planning. However, they have the benefit of ensuring that all components of an anomaly are assessed and objectively noted for management.

Table 30-5 Orbit, Mandible, Ear, Nerve, and Soft Tissue Classification System

Side: Right or Left				
Orbit	Mandible	Ear	Facial Nerve*	Soft Tissue
The midsagittal plane is defined by a vertical line from the crista galli to the anterior nasal spine, and a horizontal line is then drawn at a right angle to the midsagittal plane tangent to the supraorbital rims. Orbital features are characterized as follows:	The mandible is evaluated radiologically and categorized as follows:	External ear anomalies are categorized based on a clinical examination and standardized photographs as follows:	Facial nerve weakness is categorized based on a clinical examination as follows:	Soft tissue anomalies are categorized based on subcutaneous and apparent muscular deficiency as follows:
O ₀ : Normal orbital size and position	M ₀ : Normal mandible	E ₀ : Normal ear	N ⁷ ₀ : No facial nerve involvement	S ₀ : No obvious soft tissue or muscle deficiency
O ₁ : Abnormal orbital size	M ₁ : Mandible and glenoid fossa are small with a short ramus	E ₁ : Mild hypoplasia and cupping with all structures present	N ⁷ ₁ : Upper facial nerve involvement (temporal and zygomatic branches)	S ₁ : Minimal subcutaneous/muscle deficiency
O ₂ : Abnormal orbital position (O ₂ ¹ for superior, O ₂ ¹ for inferior)	M ₂ : Mandibular ramus short and abnormally shaped Subdivisions A and B based on the relative positions of the condyle and TMJ 2 _A : Glenoid fossa in an anatomically acceptable position with reference to the opposite TMJ (based on submental vertex views) 2 _B : TMJ inferiorly, medially, and anteriorly displaced; mandible severely hypoplastic	E ₂ : Absence of external auditory canal with variable hypoplasia of the concha	N ⁷ ₂ : Lower facial nerve involvement (buccal, mandibular, and cervical branches)	S ₂ : Moderate; between the two extremes, S ₁ and S ₃
O ₃ : Abnormal orbital size and position	M ₃ : Complete absence of ramus, glenoid fossa, and TMJ	E ₃ : Malpositioned lobule with absent auricle; lobular remnant usually inferiorly and anteriorly displaced	N ⁷ ₃ : All branches of the facial nerve affected	S ₃ : Severe soft tissue deficiency as a result of subcutaneous and muscular hypoplasia

*Other involved nerves are signified by the appropriate number in superscript (for example, N⁵ [sensory] and N¹²).
 TMJ, Temporomandibular joint.

Date: / /
 Rater: _____
 Study ID: _____

GLOBAL ASSESSMENT												
RIGHT					LEFT							
ORBIT	O0	O1	O2	O3	O4	ORBIT	O0	O1	O2	O3	O4	
S/P SURGERY	UNABLE	NORMAL	Abnormal size	Inferior orbital displacement	Superior orbital displacement	Abnormal orbital size and position	UNABLE	NORMAL	Abnormal size	Inferior orbital displacement	Superior orbital displacement	Abnormal orbital size and position
OCCLUSAL PLANE	OP0	OP1	OP2	OP3		OCCLUSAL PLANE	OP0	OP1	OP2	OP3		
S/P SURGERY	UNABLE	NO CANT	1-5 degrees	6-15 degrees	> 15 degrees	UNABLE	NO CANT	1-5 degrees	6-15 degrees	> 15 degrees		
MANDIBLE	M0	M1	M2A	M2B	M3	MANDIBLE	M0	M1	M2A	M2B	M3	
NO XRAY	UNABLE	NORMAL	Mild asx	Moderate asx	Mod-Severe asx	Severe asx	UNABLE	NORMAL	Mild asx	Moderate asx	Mod-Severe asx	Severe asx
EAR	E0	E1	E2	E3	E4	EAR	E0	E1	E2	E3	E4	
S/P SURGERY	UNABLE	NORMAL	All parts present, mild deformity	Auricle 1/2-2/3 of predicted size, not all parts present	Severely mal-formed, often peanut shaped	ANOTIA	UNABLE	NORMAL	All parts present, mild deformity	Auricle 1/2-2/3 of predicted size, not all parts present	Severely mal-formed, often peanut shaped	ANOTIA
NERVE	N0	N1	N2	N3	N4	NERVE	N0	N1	N2	N3	N4	
S/P SURGERY	UNABLE	ALL NORMAL	Brow palsy	Orbital palsy	Smile palsy	Lower lip palsy	UNABLE	ALL NORMAL	Brow palsy	Orbital palsy	Smile palsy	Lower lip palsy
SOFT TISSUE	S0	S1	S2	S3		SOFT TISSUE	S0	S1	S2	S3		
S/P SURGERY	UNABLE	NORMAL	Minimal soft tissue deficiency	Moderate soft tissue deficiency	Severe soft tissue deficiency		UNABLE	NORMAL	Minimal soft tissue deficiency	Moderate soft tissue deficiency	Severe soft tissue deficiency	
CLEFTING	C0	C1	C2			CLEFTING	C0	C1	C2			
S/P SURGERY	UNABLE	NO CLEFT	Cleft terminates medial to anterior border of masseter	Cleft terminates lateral to anterior border of masseter		UNABLE	NO CLEFT	Cleft terminates medial to anterior border of masseter	Cleft terminates lateral to anterior border of masseter			

NOTES: _____

DETAILED ASSESSMENT													
RIGHT					LEFT								
EYE	S/P SURGERY	UNABLE	NORMAL	UPPER LID COLOBOMA	LOWER LID COLOBOMA	EYE	S/P SURGERY	UNABLE	NORMAL	UPPER LID COLOBOMA	LOWER LID COLOBOMA		
	S/P SURGERY	UNABLE	NORMAL	EXOTROPIA	ESOTROPIA		S/P SURGERY	UNABLE	NORMAL	EXOTROPIA	ESOTROPIA		
	S/P SURGERY	UNABLE	NORMAL	EPIBULBAR DERMOID			S/P SURGERY	UNABLE	NORMAL	EPIBULBAR DERMOID			
EAR	E0	E1	E2	E3	E4	EAR	E0	E1	E2	E3	E4		
S/P SURGERY	UNABLE	NORMAL	All parts present, mild deformity	Auricle 1/2-2/3 of predicted size, not all parts present	Severely mal-formed, often peanut shaped	ANOTIA	UNABLE	NORMAL	All parts present, mild deformity	Auricle 1/2-2/3 of predicted size, not all parts present	Severely mal-formed, often peanut shaped	ANOTIA	
EAR CANAL	S/P SURGERY	UNABLE	NORMAL	STENOSIS	ATRESIA	EAR CANAL	S/P SURGERY	UNABLE	NORMAL	STENOSIS	ATRESIA		
TAGS	S/P SURGERY	UNABLE	NO TAGS	PREAURICULAR TAGS	FACIAL TAGS	TAGS	S/P SURGERY	UNABLE	NO TAGS	PREAURICULAR TAGS	FACIAL TAGS		
PITS	S/P SURGERY	UNABLE	NO PITS	EAR PITS	PREAURICULAR PITS	FACIAL PITS	PITS	S/P SURGERY	UNABLE	NO PITS	EAR PITS	PREAURICULAR PITS	FACIAL PITS
CLEFT	S/P SURGERY	UNABLE	NO CLEFT LIP	CLEFT LIP		CLEFT	S/P SURGERY	UNABLE	NO CLEFT LIP	CLEFT LIP			
TONGUE						TONGUE							
S/P SURGERY	UNABLE	NORMAL	Mild dysmorphisms (midline ankyloglossia or unilateral hypoplasia)	Severe dysmorphisms (lateral ankyloglossia with fusion to the mandible or severely bifid tongue)	AGLOSSIA	S/P SURGERY	UNABLE	NORMAL	Mild dysmorphisms (midline ankyloglossia or unilateral hypoplasia)	Severe dysmorphisms (lateral ankyloglossia with fusion to the mandible or severely bifid tongue)	AGLOSSIA		

NOTES: _____

Fig. 30-14 The modified pictorial OMENS-Plus classification system allows global and detailed assessments.

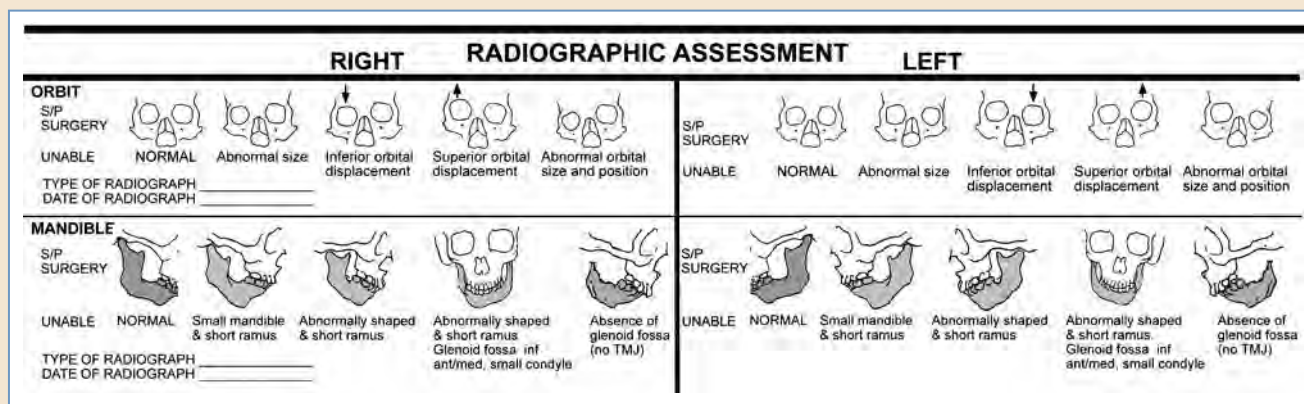


Fig. 30-14, cont'd The modified pictorial OMENS-Plus classification system also allows a radiographic assessment. (*asx*, Asymmetry; *S/P*, status post; *TMJ*, temporomandibular joint.)

MANAGEMENT OF CRANIOFACIAL MICROSOMIA

The treatment planning is individualized for every patient, given the wide spectrum of presentation. This includes not only the surgical and dental components, which are frequently emphasized, but also the needed hearing, speech, and psychological support. The intervention is ideally timed in conjunction with the growth and development of the child from infancy to adulthood. This often involves close coordination between the various interdisciplinary specialties. The speech pathologist and the plastic surgeon coordinate the need for velopharyngeal surgery. The audiologist, otolaryngologist, anaplastologist, and plastic surgeon coordinate the timing for external auricular reconstruction with placement of implants for bone conduction hearing and retention of the ear prosthesis if prosthetic rehabilitation is indicated. The plastic surgeon, orthodontist, and prosthodontist communicate to effectively plan dentoskeletal surgery. Microtia reconstruction is coordinated with the skeletal reconstruction and with soft tissue augmentation. The child and parents need to set aside time from school and work, which may be detrimentally affected by numerous appointments and surgical intervention. Thus the psychologist and the social worker are valuable members of the team, assisting families by providing support and ensuring well-coordinated care.

From a surgical perspective, broad principles and general rules guide the reconstructive sequence. The restoration of facial symmetry begins by establishing symmetry of the underlying dentoskeletal framework. Only then can soft tissue augmentation and auricular reconstruction be consistently successful. Positioning the ear appropriately with respect to the contralateral side is less likely to be satisfactory if the underlying skeletal symmetry is not established first. This fundamental principle is the ideal for patients presenting at end-stage skeletal maturity, but may need to be modified in selected cases. Many children will present in early childhood with such severe facial asymmetry that soft tissue augmentation in the form of a vascularized free flap may be required well before the child reaches skeletal maturity. Similarly, to assist children with hearing, a bone-anchored hearing device may need to be placed well before microtia reconstruction, which is dependent on the underlying skeletal symmetry. When determining the location of implant placement for a bone-anchored hearing device, surgeons need to consider future ear reconstructions; the device should not interfere with the position of later autologous or pros-

thetic reconstruction. Thus surgeons must adapt the reconstructive principles to meet the needs of each patient, the parents, and the task at hand. In many cases, the surgeon must anticipate the final form and view the reconstruction in components.

Component Facial Analysis

Patients who present with a craniofacial asymmetry require a systematic approach. Each of the components—skeletal, dental, and soft tissue—is assessed in all three anatomic planes in relationship to the other two (intrinsic spatial relationship) and in relationship to the external, three-dimensional space in which they are embedded. The natural head position (external reference frame) is noted. Knowing where the head is held in three-dimensional space is critical in understanding where to begin to establish the planes of symmetry (Fig. 30-15, *A*). Many patients with craniofacial asymmetry present with a head tilt because of ocular muscle imbalance, orbital dystopia, persistent muscular torticollis, or cervical spine abnormalities. For such patients, correction of asymmetry begins with a discussion with the patient in which the surgeon explains that the asymmetrical position of the head in three-dimensional space is to be distinguished from the internal asymmetry of the facial components. Together, they decide whether treatment will be planned with respect to the horizon, the level of the pupils, or an agreed-on external reference plane.

To determine the internal relationship of the facial components of symmetry, surgeons must establish the true facial midline, “mirror-image” plane of symmetry with which the reflection about the plane can be compared (Fig. 30-15, *B*). However, in many patients with craniofacial asymmetry involving the cranioorbital region, defining that plane can be difficult. Geometrically bisecting the interpupillary line is not always reliable. For example, in a patient with a severe craniofacial asymmetry that extends to the orbit and cranial base, the plane that bisects the interpupillary distance does not necessarily match that of a plane defined by the skull base (crista galli and sella turcica). Thus, with the more severe forms of craniofacial asymmetry, when the plane of symmetry is difficult to objectively define, clinicians should use their judgment to determine which elements can be surgically altered to an acceptable appearance.

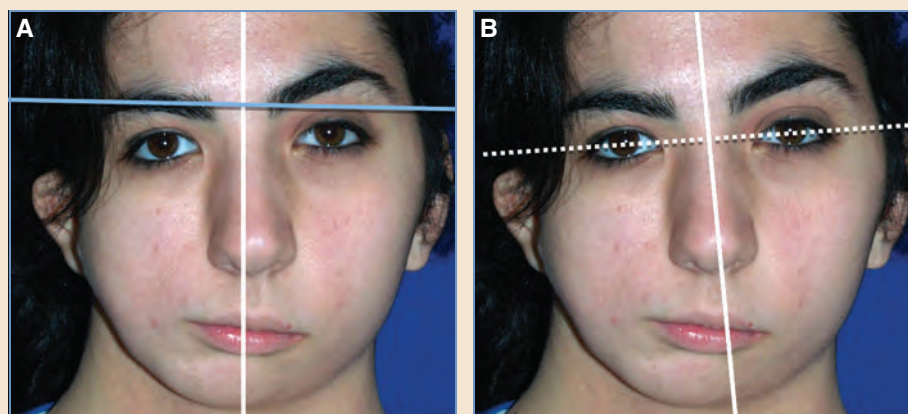


Fig. 30-15 This patient has CFM. **A**, For this photograph, she was asked to look at the distant horizon. The horizon is used as an external reference plane from which the midsagittal plane can be defined. **B**, Internal reference planes based on the interpupillary line as the horizon. The surgeon needs to decide with the patient which line of symmetry to use, if the upper midface (orbit) is not repositioned as an initial procedure.

Reyneke et al⁴⁵ provided a simplified asymmetry classification based on the chin, mandible, and maxilla to guide orthodontic and surgical management. However, once the asymmetry includes the cranioorbital structures and extends to involve the soft tissue and facial elements, such as the ears, ocular adnexa, nose, and lips, each of the components of the face must be analyzed independently for spatial position and shape. A geometric object in three-dimensional space has six degrees of freedom that determine its spatial position: three translational (delta x, delta y, and delta z) and three rotational (roll, yaw, and pitch). The definition of the shape is more complex but is intuitively represented by volume and surface topography.

In a comprehensive analysis of asymmetry, the face is anatomically broken down into its geometric components: the skeletal masses, the dental arches, the soft tissue envelope, and the individual facial elements. Each of these components is defined by its spatial position in space relative to the reflection plane or plane of symmetry and by its individual shape relative to its counterpart (Fig. 30-16).

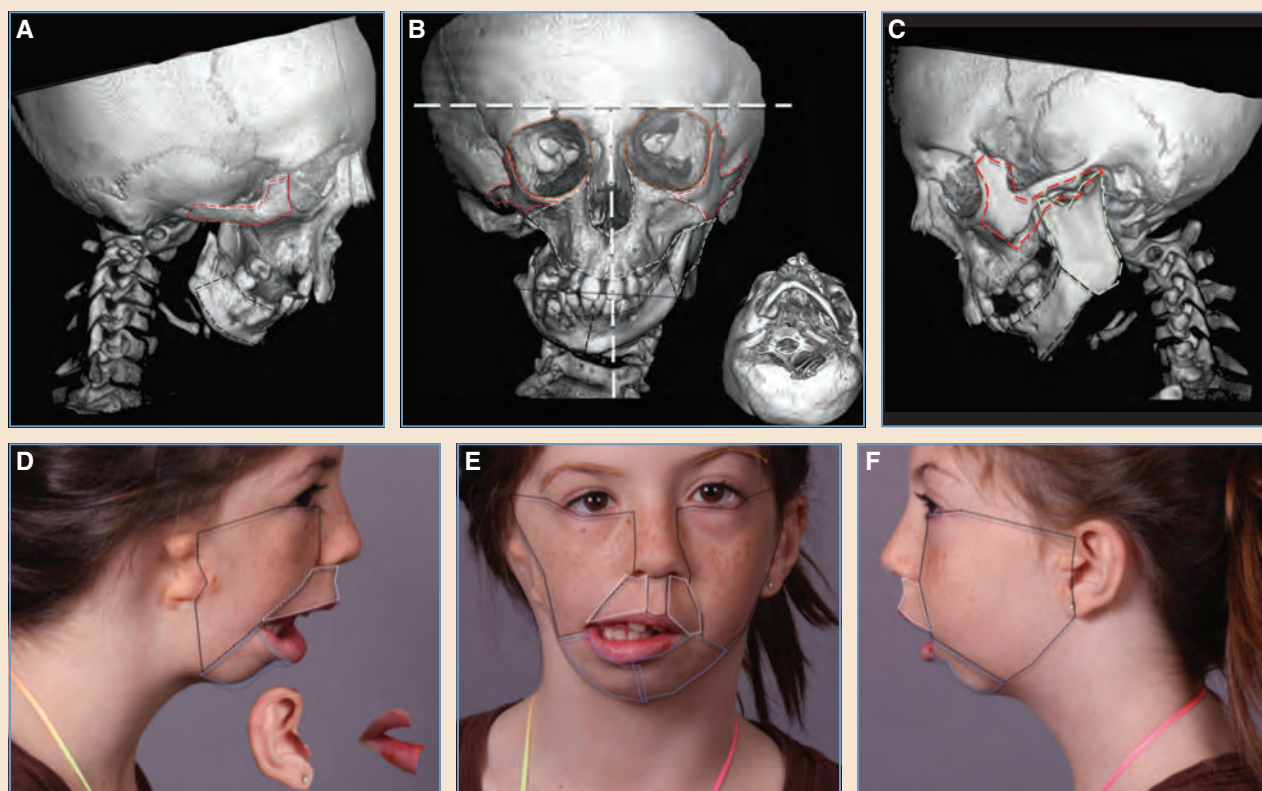


Fig. 30-16 A component analysis of facial asymmetry. **A-C**, The facial skeleton is assessed regionally based on each of the skeletal elements in terms of its three-dimensional spatial relationship to the facial midline axis and to its shape to the mirror-image counterpart. Osteotomies can be made and the elements repositioned. Autologous bone grafts are used to support the gaps. Distraction can be performed when bone is sufficient; however, vascularized free flaps are used for missing elements. Alloplastic implants may be required to restore the surface topography when repositioning the bone elements alone is not sufficient. **D-F**, This patient will require all available surgical options. The soft tissue envelope is assessed regionally, and autologous soft tissue augmentation is nearly always needed, despite restoration of symmetry to the skeletal framework. The auricular element and the macrostomia will need to be reconstructed.

The skeletal masses are the building blocks that structurally define the framework of the face. These include the frontal bone, the orbital complex, the malar-zygoma region, the maxilla, and the mandible. The mandible itself is a composite of the ramus, body, and symphysis. The dental midline, arch form, roll, yaw, and pitch of each maxillary and mandibular dental arch needs to be assessed independently. Each arch is then assessed with respect to its skeletal bases: the maxillary dental arch to the maxilla and the mandibular dental arch to the mandible. The relationship between dental arches is assessed. The thickness of the soft tissue envelope of the skeletal framework is evaluated in regional subunits, measured from the skeletal surface (volume deficiency or excess) and surface topography to achieve the desired symmetry, because correction of the un-

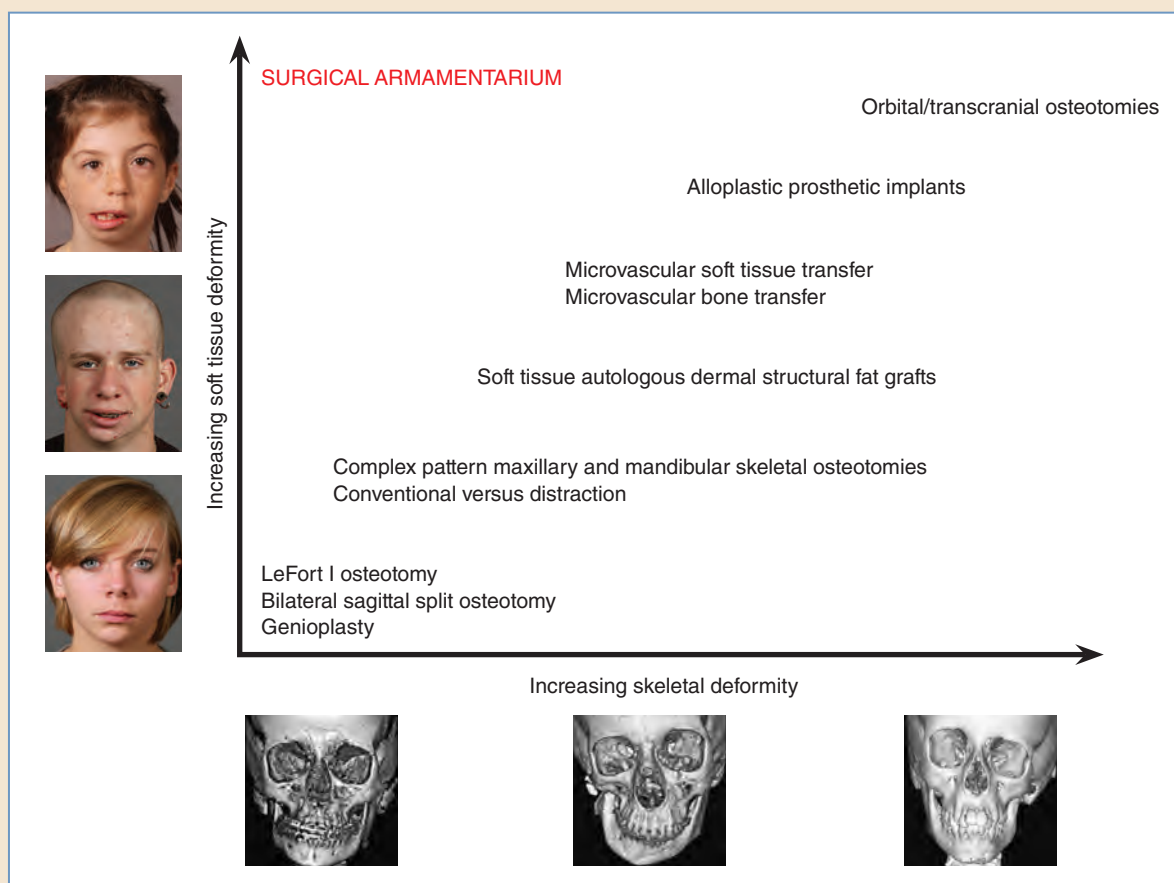


Fig. 30-17 Surgical options for skeletal and soft tissue reconstruction are tailored to the degree of severity for each component. The basic principles, regardless of the cause of asymmetry, include (1) establishing an axis of symmetry to correct roll, yaw, and pitch, (2) rearranging the existing skeleton through osteotomies and repositioning of each element, (3) including bone grafts, skeletal distraction, and microvascular bone transfer for restoration and skeletal stability, (4) refining with custom prosthetic implants to improve surface topography, (5) augmenting soft tissue through autologous grafts and microvascular tissue transfer, and (6) re-constructing missing elements such as the ear, eye, and lips.

derlying framework does not necessarily correct the soft tissue envelope. Finally, the individual facial elements that ultimately define the face are assessed for their spatial position and shape.

Thus, when a patient presents with asymmetry, every component must be carefully assessed. Only then can a systematic, multistage, reconstructive blueprint be developed. Surgeons must have within their armamentarium a large number of procedures to address each component of facial asymmetry: skeletal, soft tissue, and functional facial elements (Fig. 30-17).

Surgical Planning

Traditional surgical planning for facial skeletal and soft tissue surgery is based on clinical examination, two-dimensional photographs, lateral and frontal cephalometric radiographs, and facebow mounting of dental casts. This well-established approach has provided good results in many patients for a half century. However, the outcomes were inconsistent for patients presenting with complex facial asymmetries. The two-dimensional analyses in the orthogonal planes were difficult to integrate in surgical planning with confidence. Although the facebow transfer provides guidance, patients with CFM can have dystopic microtia resulting in auricular asymmetry. The facebow based on the ear position assumes upper facial symmetry to restore lower facial symmetry. With each step of two-dimensional planning for a three-dimensional problem—from cephalometric analysis to dental model surgery—the errors accumulated and the effects were incorporated into the occlusal splint fabrication.⁴⁶ The accuracy of the resultant occlusal splint was reduced to that of a “rough guide” only, with the final assessment of the planning based on intraoperative assessment that requires considerable judgment and experience.

Microtia reconstruction was similarly plagued with mirroring the shape and form of the normal contralateral ear and accurate positioning in three-dimensional space. Two-dimensional radiographs were used as templates. The normal ear was traced onto the film, and the templates were cut out and sterilized to be used in the operating room to construct an ear. Positioning of the ear reconstruction also involved a radiographic film. The normal ear was traced in relation to the brow, nose, and lip commissure. The reverse of the film provided the mirror image for positioning. Accuracy was difficult.

More contemporary approaches to this challenge can be performed digitally and preoperatively, allowing verification of positioning and symmetrical sizing before the actual surgery (Fig. 30-18). Two- and three-dimensional digital photography offers an efficient means of employing transparent two-dimensional replicas of the unaffected ear to position the planned reconstruction directly on the patient. However, the reconstructed visual symmetry of the pinnae may not be technically symmetrical; that is, the reconstructed ear is a mere mirror image of the unaffected ear (Fig. 30-19). In CFM patients, facial asymmetry in which horizontal lines through facial elements—eyebrows, canthi, base of the nose, and lip commissures—generally converge, indicates the need for a more intuitive approach to achieve visual symmetry rather than absolute symmetry (Fig. 30-20). An ear with absolute symmetry (that is, a mirror image) on a CFM patient with these characteristics would look far too large in comparison to the contralateral ear and would probably present challenges related to the position of the hairline. Many CFM patients have an inferiorly positioned hairline, which presents its own challenges in ear reconstruction. Perceived, visual symmetry is not absolute. Achieving the appearance of facial symmetry in these patients requires careful consideration.



Fig. 30-18 Planning techniques for autologous reconstruction. **A** and **B**, Conventional planning. **C**, Digital planning.

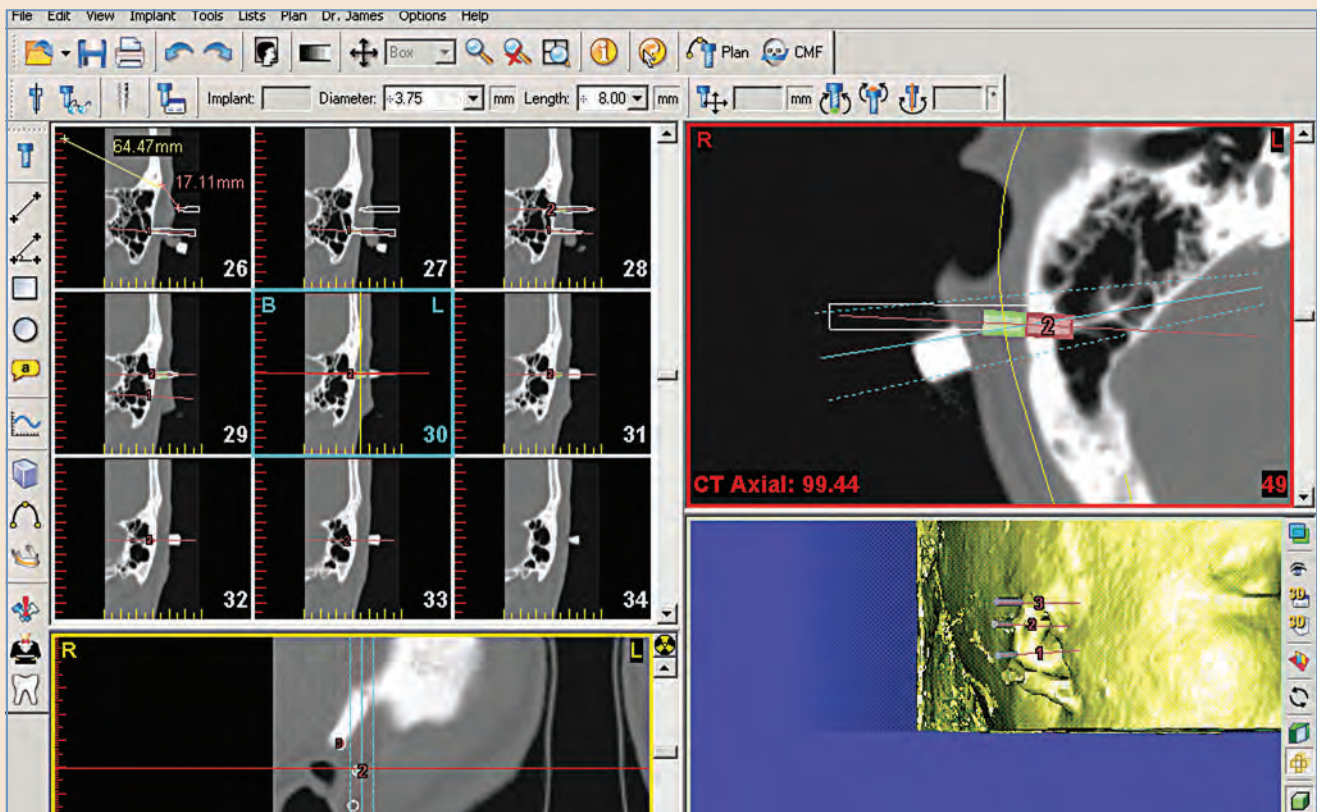


Fig. 30-19 Virtual surgical planning for accurate placement of osteointegrated implants, positioning the prosthetic ear in three-dimensional space using a mirror-image technique.

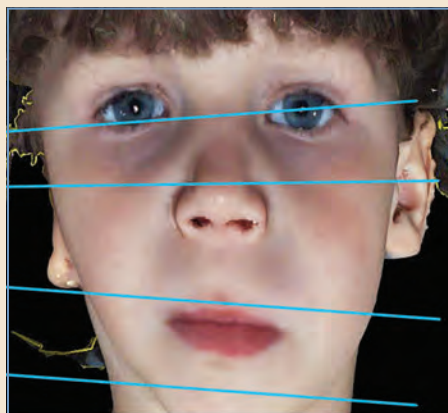


Fig. 30-20 Achieving symmetry in CFM patients is not an absolute science.

Using simple computer-based techniques and two- and three-dimensional digital photography, surgeons can make intuitive decisions toward a patient's treatment goals. An added benefit is the ability to reproduce the two- and three-dimensional photographic data into virtual planning models and surgical guides using conventional and three-dimensional printing for positioning, placement, and template shape and definition. Many of the same tools are available for planning more complex surgical interventions involving hard and soft tissues.

Currently, the software for virtual surgical planning is capable of simulating surgical procedures with ever-increasing user applicability, from the imaging research laboratory to routine daily clinical application.⁴⁷⁻⁴⁹ The facial skeleton is treated as a solid, three-dimensional object in virtual space that can be digitally manipulated. Osteotomy planes can be defined, and the three-dimensional facial skeletal object can be digitally sectioned into two separate solid objects, allowing each to be moved independent of the other. The software treats the CT data set as solid objects on which Boolean algebra can be performed. Three-dimensional visual objects can be added and subtracted digitally. Using the registration and superimposition technique, it is possible to build an integrated, composite model of the dentition, based on both volumetric information from CT or cone-beam CT and surface information from laser or optical scans with greater clinical accuracy. Surgery can be planned in three-dimensional space instead of in planar projection, two-dimensional cephalometric films. The complex facial skeletal asymmetries can be addressed. Moreover, the conventional inaccuracies and time-consuming laboratory process of facebow transfer, plaster mounting of dental casts, sectioning, repositioning, and remounting of dental casts are eliminated. Surgeons can execute various surgical options to optimize outcomes. Regions of interference during the skeletal movement can be assessed and detected (for example, in patients with maxillary impaction and mandibular rotation), the dental roots can be visualized within a skeletal framework for interdental osteotomies, and the course of the inferior alveolar nerve can be visualized for mandibular osteotomies. The mirroring function of the software provides a more accurate template for microtia reconstruction, compared with the conventional use of two-dimensional radiographic templates. The templates to construct an ear and the positioning can be three-dimensionally printed.

Component Reconstruction

The sequence of reconstruction (beginning with the skeletal framework, the overlying soft tissue, and followed by the various specialized components) is described in the following section. Each component is integrated either in parallel or sequentially with the child's growth and development, functional needs, psychosocial concerns, and aesthetic goals (see Table 30-6).

Reconstruction of the Skeletal Framework***Cranioorbital Reconstruction***

In a small percentage of patients with CFM who present with frontoorbital involvement, establishing symmetry of the lower facial region (maxillary-mandibular) is dependent on establishing upper facial symmetry. When the patient is in childhood or early adolescence, a decision is made with the patient and family whether to address orbital dystopia through transcranial "box" orbital osteotomies. Currently available three-dimensional surgical planning software is invaluable in assisting surgeons in positioning the orbit using the mirror-image function. The technical details of repositioning and orbital expansion osteotomies are well described in the literature.^{50,51}

Maxillary-Mandibular Dental Reconstruction

Established surgical principles involve complete disassembly and reassembly of the maxillofacial skeleton, carefully designed osteotomies, repositioning and fixation of the bony elements to their desired position, and rigid skeletal fixation systems supported by bone grafts.⁵² These classical procedures, the LeFort I osteotomy, bilateral sagittal split osteotomy, and genioplasty, are described in detail in Chapter 27. For patients with CFM, a classic LeFort I osteotomy pattern is modified to account for the asymmetry to achieve a desired result (Fig. 30-21). The osteotomy on the uninvolved side of the face may follow the classic downward-sloping osteotomy line through the maxilla, and on the deficient side, the osteotomy may be upward sloping to include a portion of the zygoma. The mandibular osteotomies will also be asymmetrical to achieve a more symmetrical outcome. For patients who have a relative degree of symmetry, surgeons need only address the rotation of the maxillary-mandibular complex in the sagittal plane to achieve a proper pitch or occlusal cant. The complexity with CFM patients involves a rotational element not only in the sagittal plane but also in the coronal plane (roll or cant) and the transverse plane (yaw). Therefore repositioning with six degrees of freedom adds a significant level of complexity to obtain the desired outcome. Moreover, the desired asymmetry has anatomic limitations. The impact of mandibular rotation on the condyle within the glenoid fossa can be significant. Surgeons should be concerned about internal derangement of TMJ function. The mandibular shape itself is abnormal, and achieving symmetry is limited through osteotomies and repositioning alone.

These procedures are performed presuming bony material is sufficient to rearrange the skeletal framework to achieve the surgical goals, but this is rarely the case in CFM patients. The exception to this rule is in Pruzansky grade I patients, in whom asymmetry is minimal, although clinically evident. If skeletal stability is a concern because of the amount of desired advancement (bone length) and excessive soft tissue and muscular forces contributing to relapse, skeletal distraction is added to the surgeon's armamentarium. Classic orthognathic procedures and distraction osteogenesis procedures use the same osteotomy pattern; the fundamental difference is the rate of displacement of the bone segments. With classic orthognathic procedures, the immediate final position of the bone segment is subject to strong relapsing forces and needs to be supported by skeletal fixation and bone grafting of the osteotomy gap. With distraction osteogenesis, the slow rate of displacement in the range of 1.0 mm/day allows gradual accommodation of the soft tissue and muscle matrix in which the skeletal framework is embedded. The biologic principles and the clinical application have been well described since its introduction in 1990.⁵³⁻⁵⁵ Distrac-

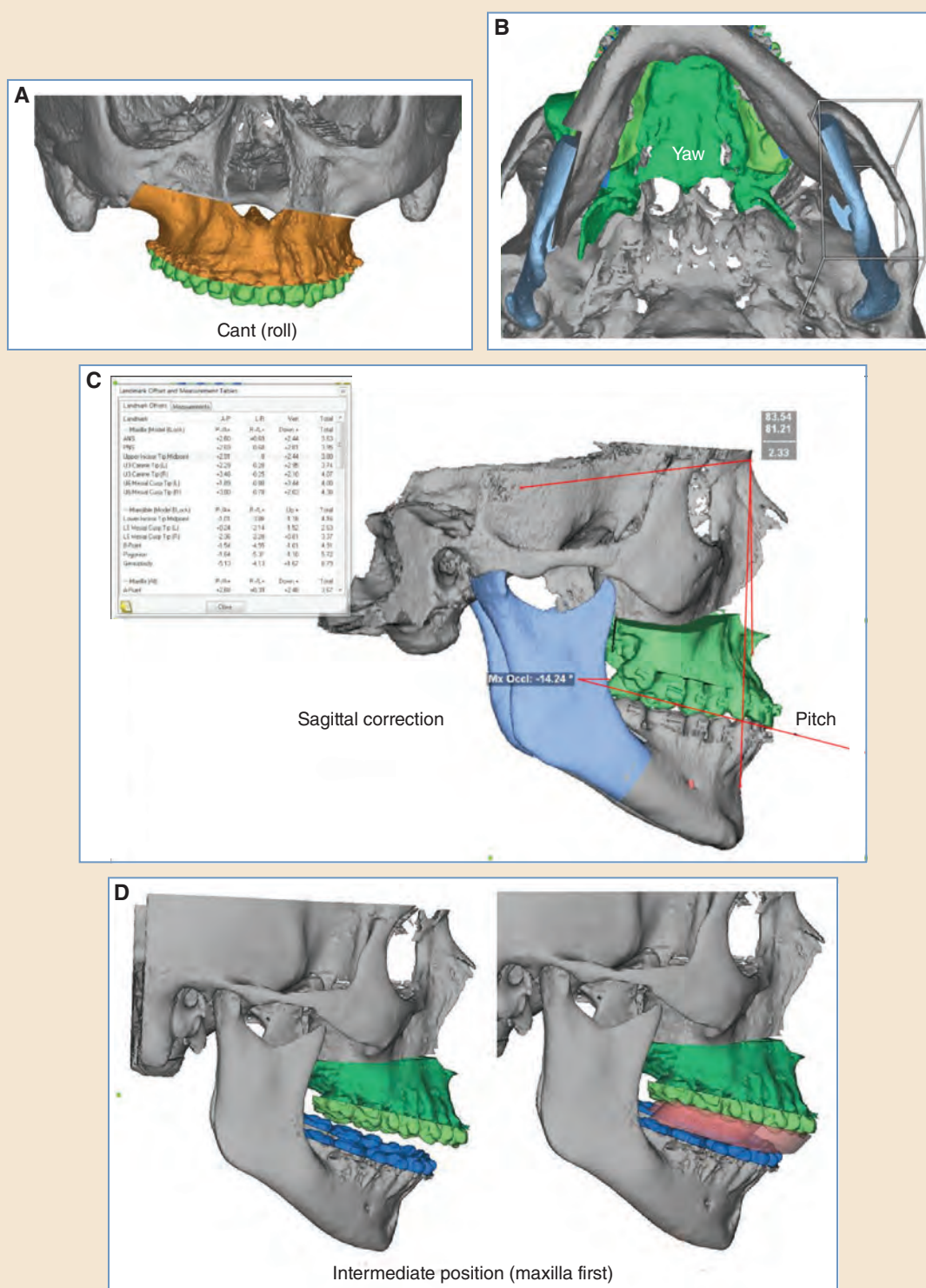


Fig. 30-21 Virtual surgical planning of orthognathic surgery (model surgery). **A**, An asymmetrical LeFort I osteotomy tailored to a patient's deformity with the cant (roll). **B**, Yaw correction after a double-jaw surgery and assessment of interference at the bilateral sagittal splitting osteotomy junction. **C**, Sagittal correction with dynamic cephalometric analysis. **D**, An intermediate position with maxillary surgery first and design of the intermediate splint to be fabricated for intraoperative positioning of the maxilla relative to the mandible (see the accompanying video).

tion osteogenesis is an important surgical tool in mandibles classified as Pruzansky grade II, in which bone stock is insufficient for the classic orthognathic procedures but sufficient for distraction. With mandibular distraction alone, an open bite is created on the involved side, allowing the maxilla to be leveled with orthodontic appliances (Fig. 30-22). With simultaneous maxillary distraction driven by mandibular distraction, both jaws can be leveled to the extent limited only by the vector control of the distraction device (Fig. 30-23). In severe cases, the desired symmetry may be difficult to obtain through distraction alone, particularly when multiple vectors for lengthening are required. In these cases, the goals of distraction may be to achieve satisfactory maxillary position and sufficient mandibular length to facilitate a classic orthognathic procedure as a secondary procedure, offering surgeons more control in achieving symmetry (Fig. 30-24).



Fig. 30-22 This patient had left Kaban-Pruzansky type IIA CFM. Distraction osteogenesis was performed for mandibular lengthening with an external device. **A** and **B**, Before the distraction. **C**, During mandibular distraction osteogenesis. **D** and **E**, The postoperative result.

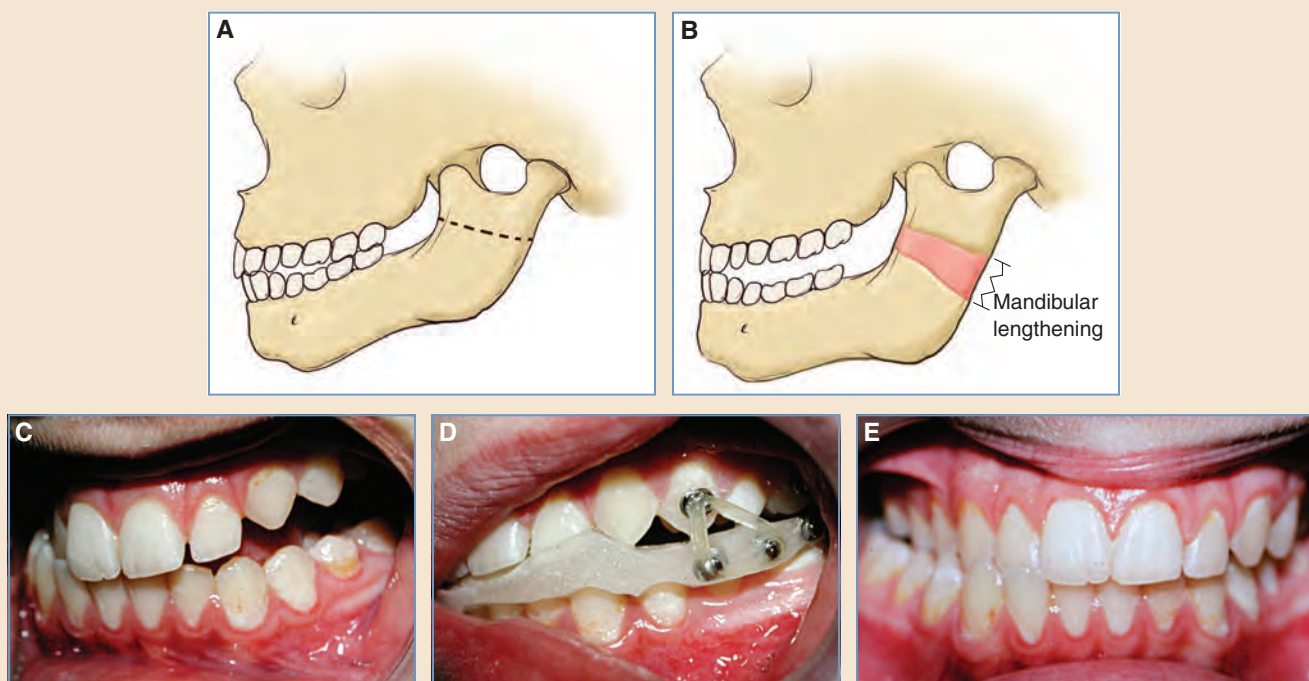


Fig. 30-23 A and B, Management of an occlusal open bite after left mandibular lengthening. C, After distraction, the resultant crossbite was evident. D, Orthodontic management. E, The contralateral right crossbite could have been corrected with a simultaneous palatal expansion. This occurs because of the inability to fully control the multivector direction with a single-vector distraction device.

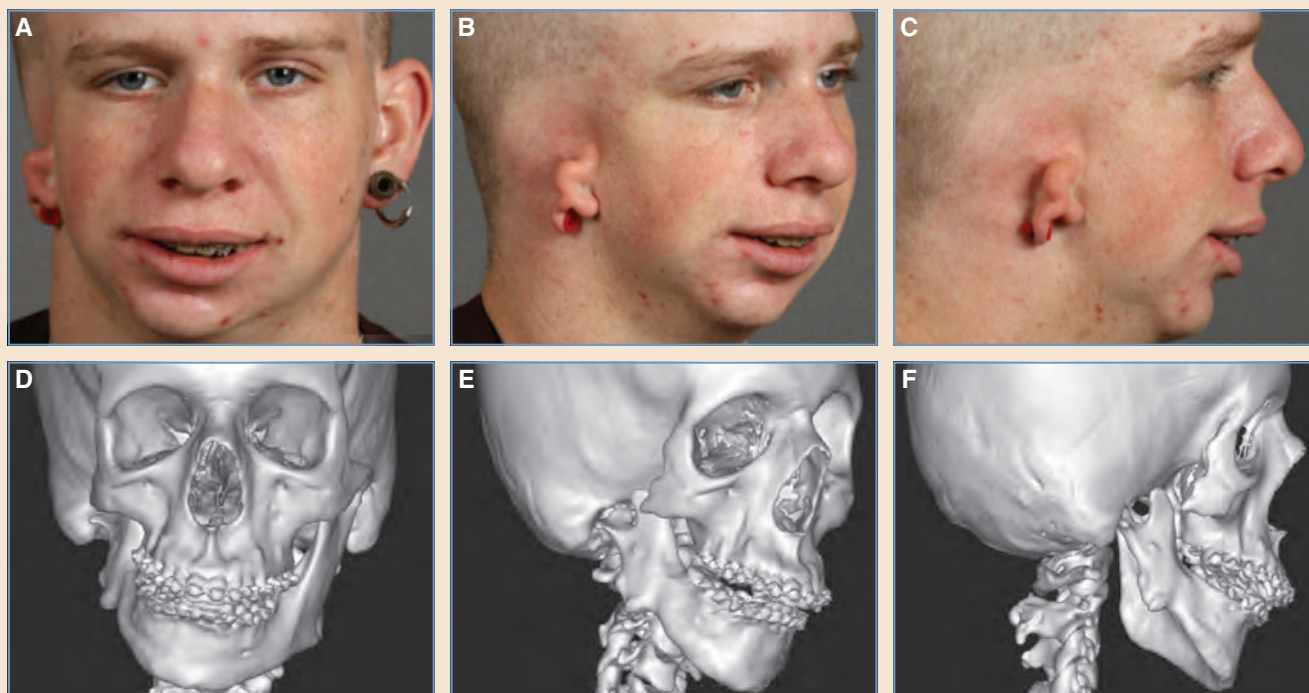


Fig. 30-24 This patient had right CFM (Kaban-Pruzansky type IIB). A-F, Preoperative clinical images and CT scans showed the underlying skeletal anatomy.

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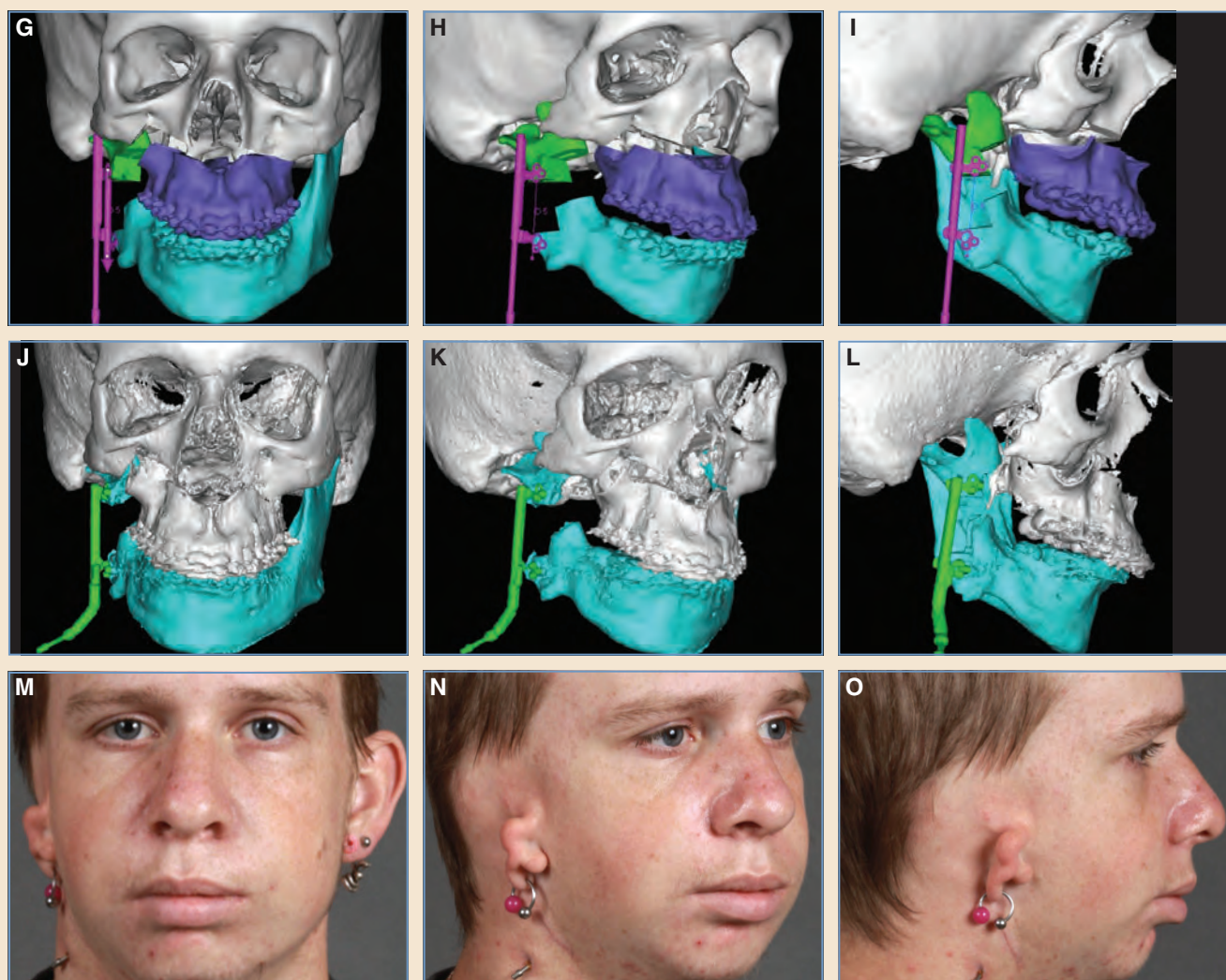


Fig. 30-24, cont'd G-L, Surgical simulation of simultaneous maxillary and mandibular distraction. The virtual simulation offered surgeons an opportunity to optimize the vector of distraction. However, the true kinematics of the right temporomandibular joint could not be reproduced. M-O, The outcome at 8 weeks. The internal device was maintained until consolidation.

Distraction osteogenesis offers the benefit of harnessing patients' normal growth and development with earlier intervention. In patients with CFM, the benefits of an earlier mandibular distraction-lengthening procedure include improvements in the airway by repositioning the tongue base and lessening the secondary impact on the midfacial skeleton by removing the anatomic restriction of the mandible.⁵⁶ Nevertheless, surgeons should inform patients and their parents that, after these early interventions, further mandibular lengthening will be needed later in childhood, supported by orthodontic management, and conventional orthognathic surgery will be required in adolescence. There is a definite role for early, interceptive orthodontics, combined when needed with surgery, which can reduce the degree of asymmetry until more definitive skeletal procedures can be performed at maturity.

When bone is insufficient for distraction (for example, a Pruzansky grade III mandible), then the alternatives are nonvascularized bone grafts (costochondral rib grafts or iliac bone grafts) and vascularized bone transfer (fibula, scapula, or radius) through microsurgery techniques^{57,58} (Fig. 30-25). Once sufficient bone stock is present (that is, a Pruzansky grade III mandible is converted to a Pruzansky grade II mandible), distraction osteogenesis and classic orthognathic procedures can be completed to reduce the skeletal asymmetry.

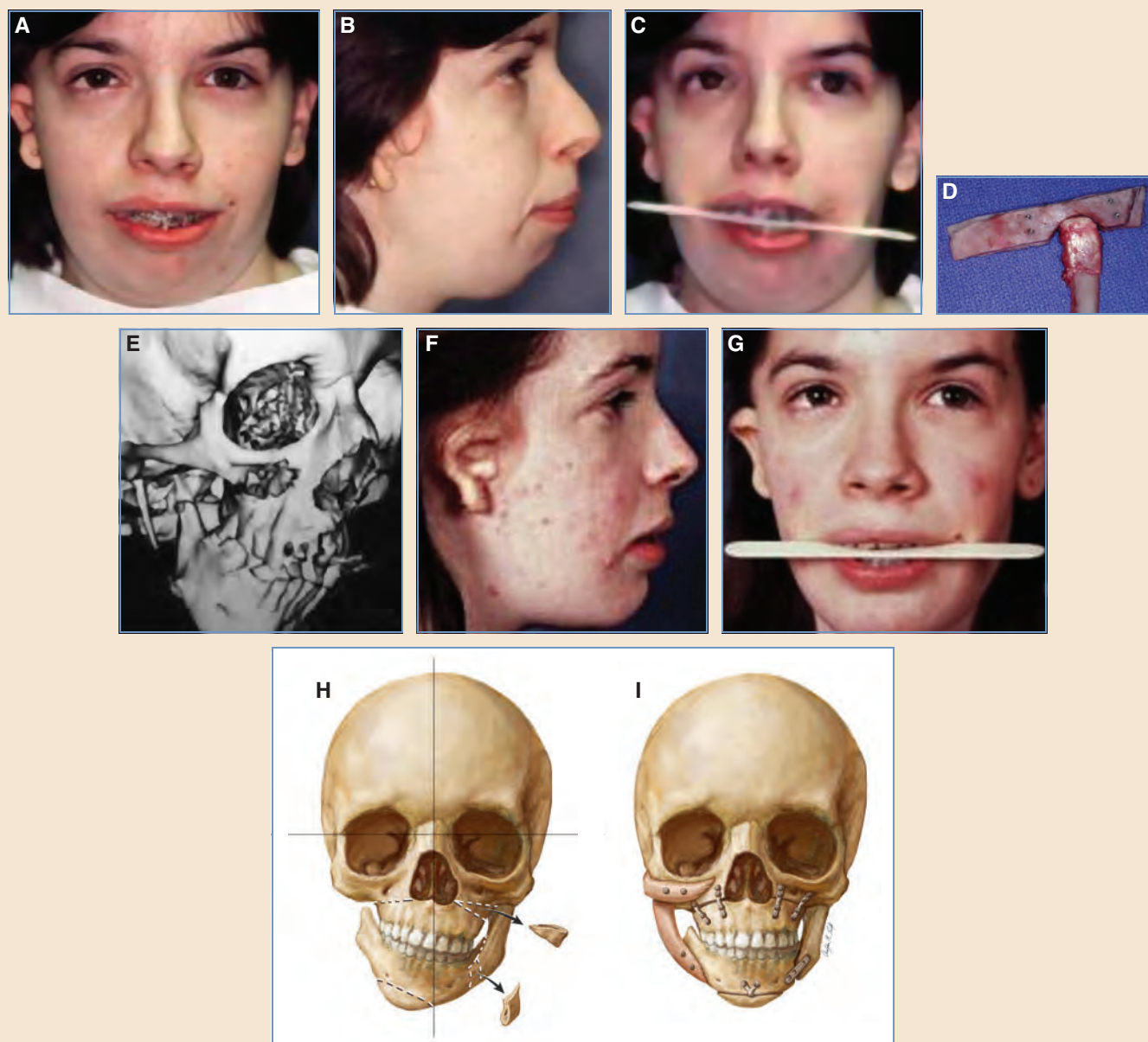


Fig. 30-25 This patient with right CFM (Pruzansky grade III) underwent an asymmetrical LeFort I osteotomy, a left sagittal split osteotomy of the ramus, and a right rib graft and fibular reconstruction. **A-C**, Preoperative views showed occlusal cant and right-sided maxillary and mandibular deficiency. **D**, A fibula and rib graft for right mandibular reconstruction. **E**, A three-dimensional CT scan demonstrated the right-sided deficiency. **F** and **G**, Postoperative views show improved symmetry and correction of the occlusal plane. **H** and **I**, The reconstruction included asymmetrical osteotomies and graft placement.

Even with carefully tailored osteotomies and repositioning of the existing skeletal framework, residual asymmetry can result because of the abnormal shape of the bony contours of the mandible and the maxilla (Fig. 30-26). Skeletal contouring procedures are important to achieve the final symmetry. Autologous bone grafts and custom-designed, mirror-image alloplastic implants are required to restore the shape or the surface topography of the involved skeletal element, primarily the mandibular border and the maxillary-zygoma region.⁴⁹

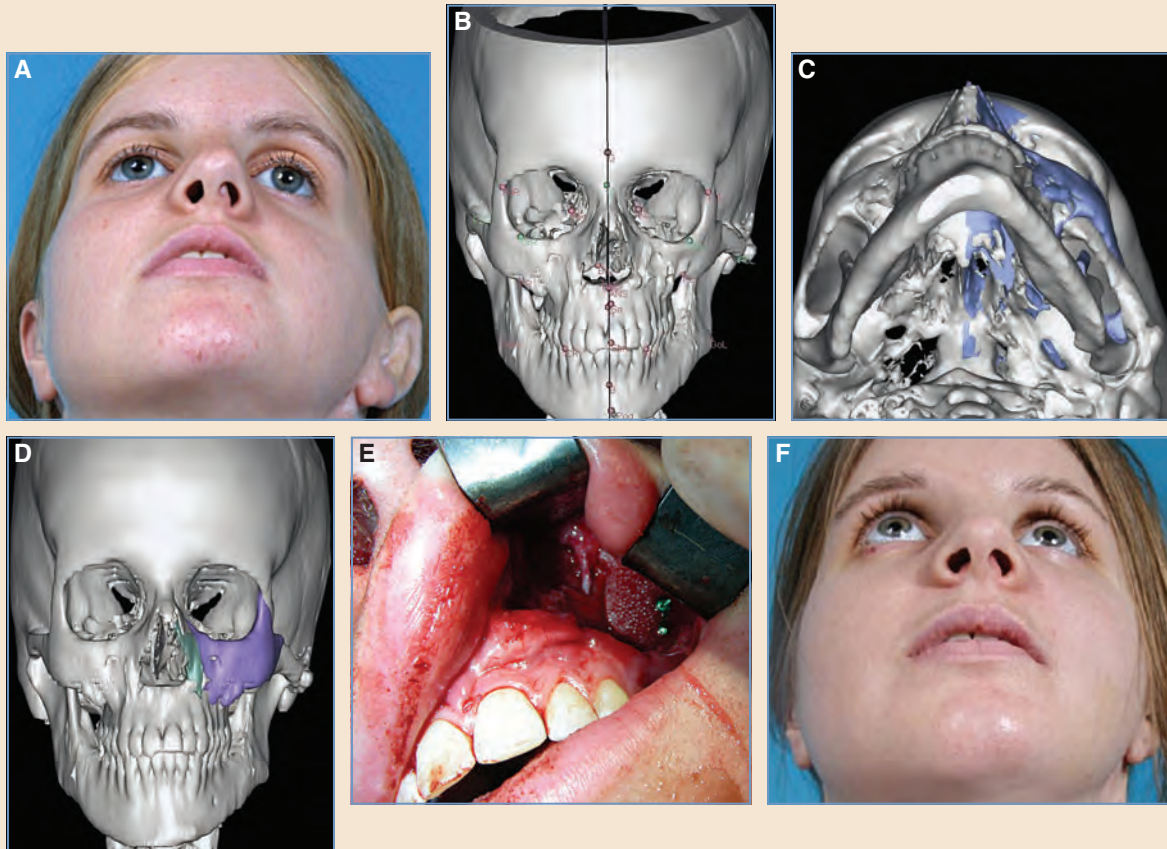


Fig. 30-26 This patient had left CFM after a LeFort I osteotomy and a bilateral sagittal splitting osteotomy. **A**, A worm's-eye view showed residual midface asymmetry. **B**, A three-dimensional CT scan revealed residual zygomatic deficiency. **C** and **D**, Three-dimensional CT planning for correction of bony asymmetry with an alloplastic implant. **E**, Intraoperative placement of a porous polyethylene implant. **F**, The postoperative result.

Soft Tissue Reconstruction

Once the skeletal form is addressed, then soft tissue asymmetry of the overlying skin envelope can be corrected through a microsurgical vascularized adipofascial tissue transfer, nonvascularized dermal fat grafting, and structural fat grafting. When a large volume is required, a microvascular tissue transfer provides a good solution for patients with severe deficiency.⁵⁹ The choices include scapula, parascapula, anterolateral thigh, and deep inferior epigastric perforator flaps. However, tailoring the augmentation to specific facial regions is difficult, and many will require subsequent debulking and refinement procedures. Moreover, the appearance of the donor site needs to be carefully discussed. In many patients whose soft tissue deficiency is mild to moderate, nonvascularized dermal fat grafting is an appropriate choice. Some degree of volumetric resorption almost always occurs with nonvascularized grafts; therefore the augmentation is overcorrected generally by 20% to 25%, and some will require additional grafting. In recent years the introduction of serial, staged structural fat grafting has revolutionized the soft tissue asymmetry management in many patients. Fat is harvested from the abdominal and flank regions, purified, and infiltrated in small aliquots (0.1 cc) in a highly controlled manner in multiple planes, offering the surgeon a degree of precision that cannot be achieved by other approaches. As with dermal fat grafting, some resorption is inevitable, and structural fat grafting is generally repeated until the desired level of symmetry is achieved.⁶⁰⁻⁶²

Auricular Reconstruction

For severe microtia or anotia requiring total ear reconstruction with autologous cartilage, it is preferable to wait until patients are 10 years of age.⁶³ This ensures adequate costochondral cartilage is available and minimizes the resultant contour deficit at the chest donor site. Chapter 31 provides a more complete discussion of autologous total ear reconstruction. CFM patients present a number of unique challenges involving surgical planning, positioning of the reconstruction, and skin coverage. These issues are presented here.

Auricular Dystopia

With increasing severity of CFM, deficiencies of the underlying facial and cranial base skeleton can result in anterior, inferior, and medial displacement of the microtic vestige. The reconstructive plan for this displacement, known as *auricular dystopia*, should include restoration of a normal ear *position* and normal ear *form*. In patients with significant displacement, this may necessitate repositioning the dystopic vestige as a separate, preliminary operation before placing the auricular cartilage framework to maximize its blood supply for further movement at the later operation. The dystopic vestige is usually repositioned by Z-plasty, although it may also be transposed posteriorly on the leading edge of a cheek flap or as an island flap. This repositioning allows placement of the vestige closer to its ultimate position and increases the distance between the ear and the lateral canthus, both of which contribute to a less conspicuous deformity. This can be performed in early childhood, several years before the main reconstruction, without adverse effects⁶⁴ (Fig. 30-27).

In a few patients, the dystopic vestige can be associated with a patent external auditory meatus and canal. These structures and the adjacent facial nerve may limit the amount of available movement. The cartilaginous canal can be dissected sufficiently to allow pivoting around its connection with the bony canal, but the two segments must be kept in continuity. The superficial temporal and postauricular arteries should be preserved for future fascial flap transpositions. Preoperative imaging with CT or MRI can be helpful in determining the course of the canal, facial nerve, and vessels.

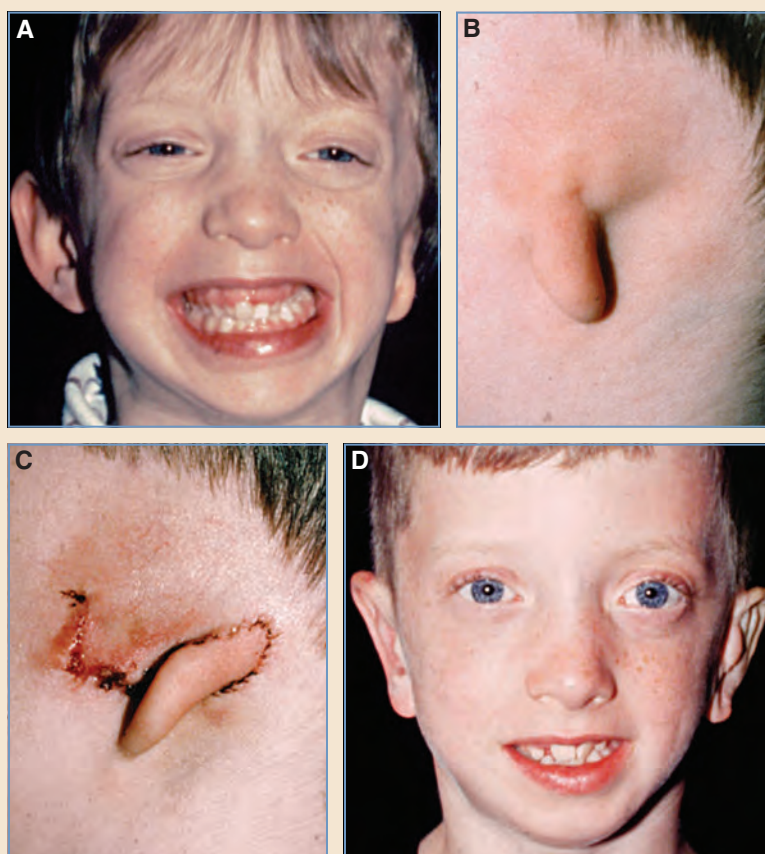


Fig. 30-27 This patient underwent transposition of a dystopic vestige. **A** and **B**, Preoperative views of the dystopic lobular remnant. **C**, Transposition of the dystopic remnant by a Z-plasty technique. **D**, The final result shows excellent symmetry.

Skeletal Hypoplasia

CFM patients with a hypoplastic temporal bone or cranial base may present an inadequate foundation for the ear, and reconstructive efforts that do not account for this are unlikely to be successful. The reconstructed auricle may be of appropriate shape and positioned appropriately in the transverse and coronal planes, but inadequate bony support leads to a reconstructed ear that remains *medially* displaced, particularly at the lower pole. The result is an ear that appears to have been sheared off the side of the head (Fig. 30-28, *A* and *B*). To address this, it is necessary to provide additional support for the cartilage framework when it is placed. In patients with mild skeletal hypoplasia, an additional block of cartilage can be placed under the main construct to provide the necessary projection, whereas in severe cases, a cantilevered cranial bone graft from the temporal bone may be necessary^{64,65} (Fig. 30-28, *D* and *E*).

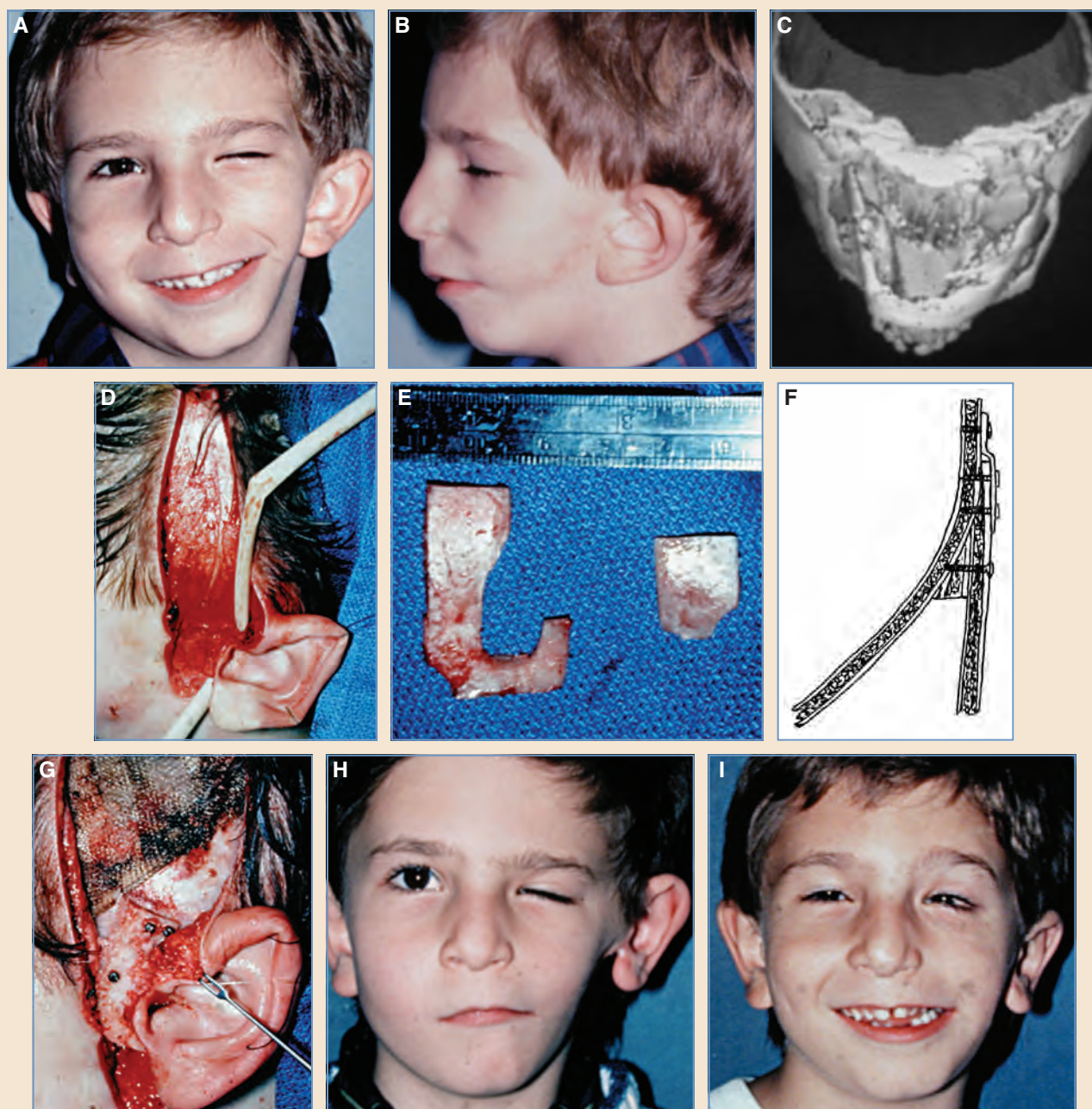


Fig. 30-28 This patient had temporal bone hypoplasia associated with CFM. **A** and **B**, His ear was normal but malpositioned because of an underlying skeletal deficiency. **C**, CT reconstruction revealed a steeper angle of the temporal bone related to the craniofacial hypoplasia. **D**, Intraoperative exposure for cantilever bone graft placement. The external auditory canal was identified, and dissection was carried circumferentially to the level of the temporal bone. **E**, A cantilever bone graft and an additional bone strut. **F**, The arrangement of the grafts on the temporal bone. **G**, Grafts were secured to the temporal bone, encircling the canal. **H**, An early postoperative view, before mandibular reconstruction and ptosis repair. **I**, The final result.

Skin Coverage

Surgeons should be aware of the ultimate position of the ear when planning reconstruction. Many patients with CFM have a low hairline on the affected side. If the construct will require placement under mostly hair-bearing skin, then it may be more prudent to use a fascial flap with a skin graft to provide hair-free skin coverage in these cases (Fig. 30-29). Similarly, scarring from previous attempts at ear reconstruction, mandibular distraction, or bone-anchored hearing aid (BAHA) placement can potentially preclude placement of the construct under a healthy skin envelope, and instead necessitate the use of a fascial flap with a skin graft.^{66,67} For this reason, when possible, these earlier interventions are planned with the auricular reconstruction in mind, and placement of the incisions is adjusted accordingly.

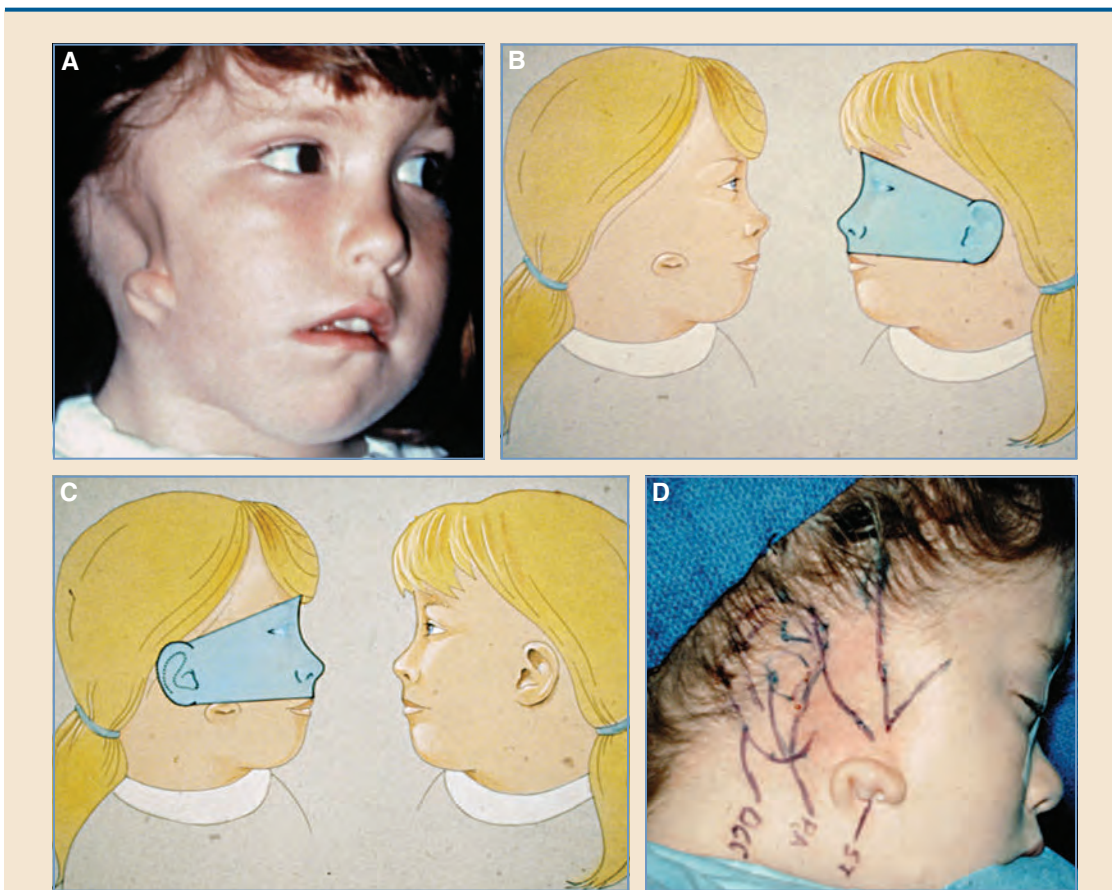


Fig. 30-29 Auricular reconstruction with rib cartilage, a temporoparietal fascial flap, and skin graft coverage. The use of a fascial flap and skin graft was preferred in this case. The patient's low hairline would have resulted in hair-bearing skin covering a large portion of the reconstructed ear. **A**, A preoperative view showed right microtia with an anteroinferiorly displaced vestige. **B** and **C**, A clear template from the contralateral side helped to determine the correct position for the reconstructed ear. **D**, Operative markings confirmed the low-set hairline. Approximately half of the reconstructed ear would be placed under hair-bearing skin. The dystopic vestige was also repositioned at this stage.

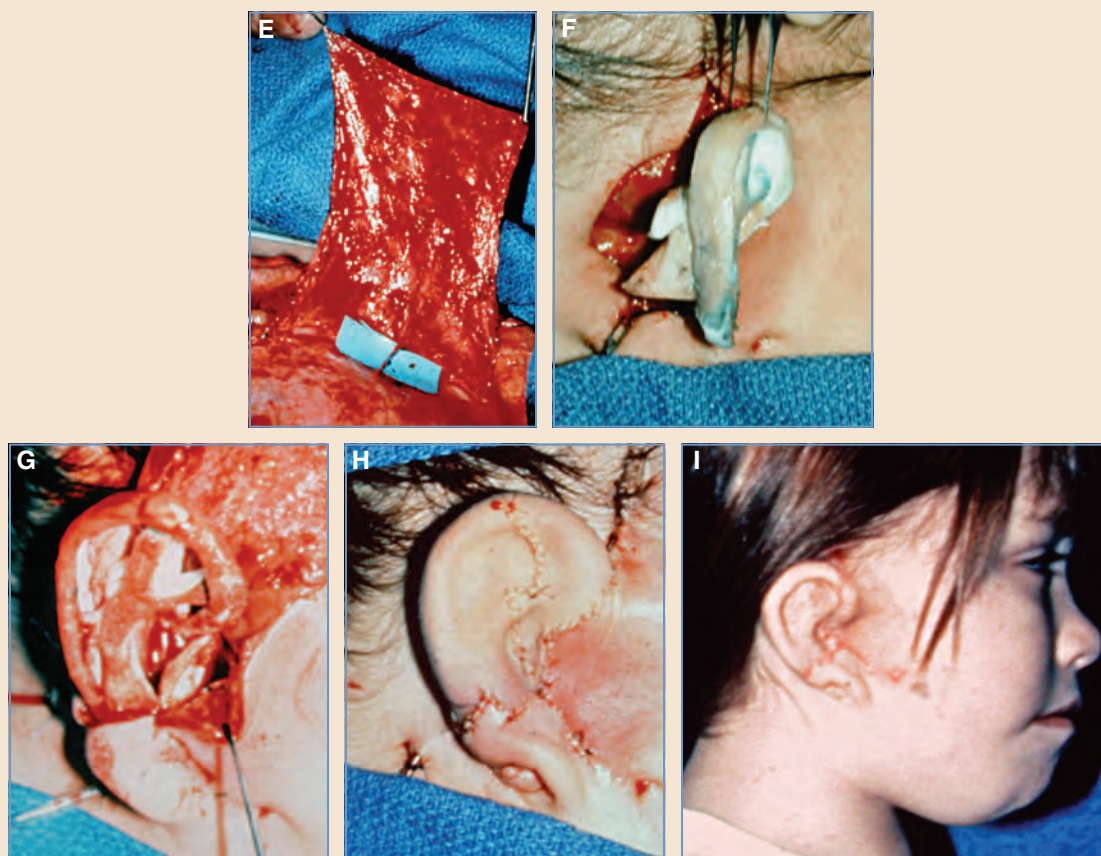


Fig. 30-29, cont'd E, Harvest of a pedicled temporoparietal fascial flap, over which a skin graft can be placed. The superficial temporal artery is isolated. F, Placement of the rib cartilage framework. In this case, an additional cartilage block was placed behind the framework to compensate for the temporal bone deficiency. G and H, The auricular framework was positioned and covered with a full-thickness skin graft. I, The completed reconstruction.

Alloplastic Reconstruction

Alloplastic auricular reconstruction is an alternative to autologous reconstruction that involves placing a biocompatible porous polyethylene implant (Medpor, Stryker) under a vascularized temporoparietal fascial flap. The flap is covered with an anteriorly based skin flap and skin graft. Reinisch and Lewin⁶⁸ pioneered the technique. It provides a secondary option for patients who do not desire, or are not suitable candidates for, autologous reconstruction. The reported benefits include a shorter procedure with no donor-site pain or morbidity, single-stage reconstruction, and performing the reconstruction before patients enter school. With the use of custom implants, surgeons can better mimic the intricate features of the contralateral ear. However, the technique has disadvantages similar to those of other implant-based procedures, including infection and implant extrusion. Any exposure or threatened extrusion of the implant necessitates another operation, because the presence of a foreign body impedes healing of the overlying tissue. Salvage procedures are difficult. Although the rate of reported implant fracture is as high as 5% in patients undergoing atresia repair, the long-term failure rate is higher, as with any foreign body placement, and is unpredictable.^{68,69}

Auricular Prosthesis

Auricular prostheses can be custom fabricated to match a patient's contralateral ear and may provide the most symmetrical result, compared with all other reconstructive options (Fig. 30-30). The indications for auricular prosthesis in CFM patients include patient preference, poor operative candidates, salvage of an unsuccessful autologous reconstruction, and severely compromised tissue.⁷⁰ The benefits of prosthetic reconstruction over other means of reconstruction include minimal morbidity, excellent aesthetic results, and the ability to salvage with other types of reconstruction. The disadvantages include not using patients' own tissue, possible social embarrassment if the prosthesis becomes dislodged, and the need for regular replacement, which involves added expense and requires access to an appropriate provider.⁷¹ Table 30-6 provides a review of component reconstruction.

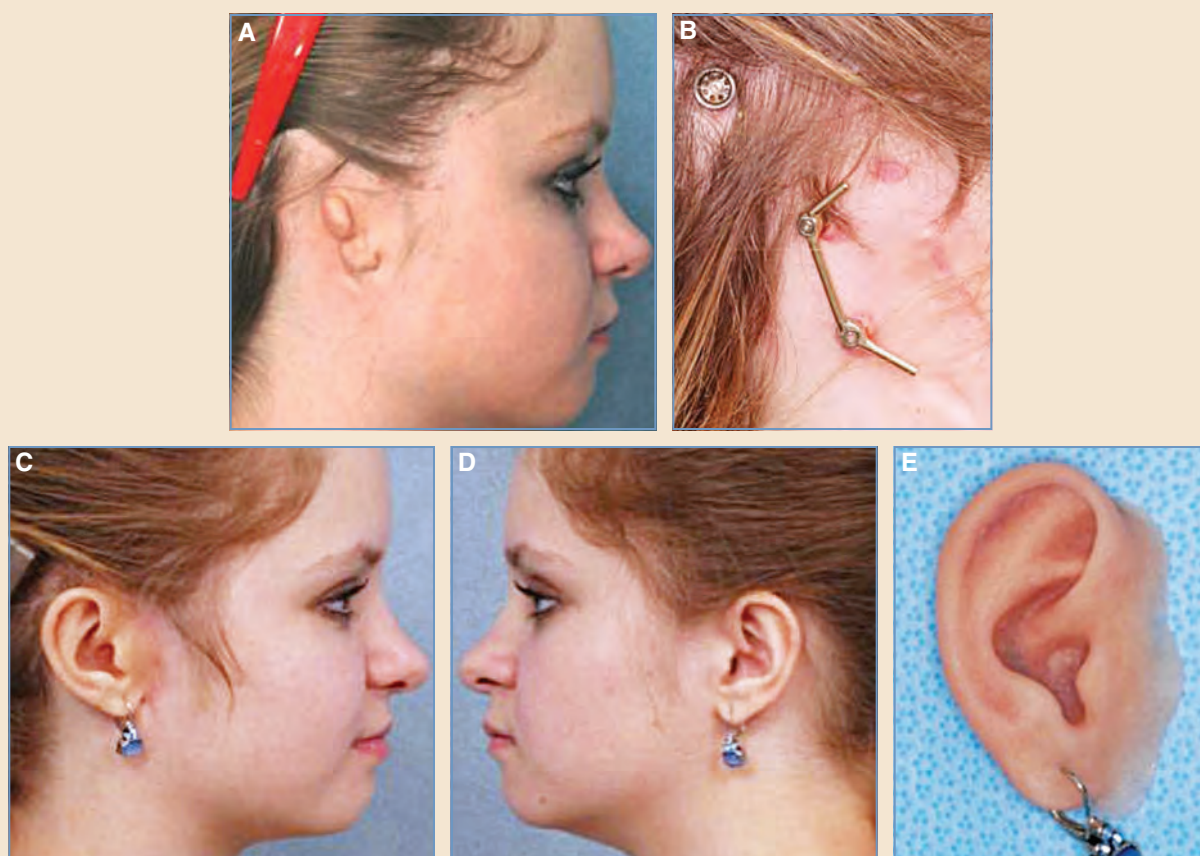


Fig. 30-30 This patient had right microtia and elected to have osteointegrated prosthetic ear reconstruction with simultaneous placement of a BAHA. **A**, Right anotia. **B**, A bar-and-clip retention device, anchored with osteointegrated titanium screws. **C** and **D**, Postoperatively, the right auricular prosthesis has excellent symmetry with the contralateral ear. **E**, The auricular prosthesis.

Table 30-6 Component Reconstruction

Age	Dentoskeleton	Speech	Ear	Mouth	Soft Tissue	Nerve
Infancy	Type II: Mandibular distraction Type III: Rib graft		Removal of preauricular remnants Lobule transposition	Macrostomia repair		
Early childhood		VPI surgery				
Late childhood	Type II: Mandibular distraction with orthodontic support Type III: Rib graft/vascularized fibula Growth modification phase I orthodontic management		Bone-anchored hearing aid		Dermal fat grafting Vascularized soft tissue flap Autologous fat grafting	Facial nerve reanimation Gold weight Lower lip botulinum toxin and myectomy
Early adolescence			Staged autologous microtia reconstruction			
Late adolescence to adulthood	Comprehensive phase II orthodontic management Type I: LeFort I, BSSO versus inverted L, genioplasty Type II: Simultaneous maxillary-mandibular distraction Type III: Rib graft/vascularized fibula Implant-retained dental prosthesis		Prosthetic ear reconstruction (failed microtia)			

Reconstruction must be tailored to each patient's needs and the staging of various components optimized for anatomic and functional benefit. *BSSO*, Bilateral sagittal split osteotomy; *VPI*, velopharyngeal insufficiency.

The prosthesis is made of silicone, and is color matched to the patient's skin for the best possible result (Fig. 30-31). The prosthesis is attached using a skin adhesive or by surgical placement of osteointegrated titanium screws into the temporal bone. With successful osteointegration, component retention systems can be designed using a bar-and-clip design or magnet retention. The benefits of osteointegrated implants include more reliable retention, ease of positioning and placement, and a longer expected lifespan of the prosthesis. Disadvantages of prosthetic reconstruction include the requirement for one or two surgeries for implant placement and abutment connection, respectively; design challenges associated with placing implants in inadequate bone where skeletal anatomy may be anomalous; aging of the silicone, as noted by discoloration and tearing of the prosthesis; and the patient's required commitment to daily hygiene maintenance of the periabutment tissue and prosthesis⁷² (Table 30-7). Prosthetic devices are custom made

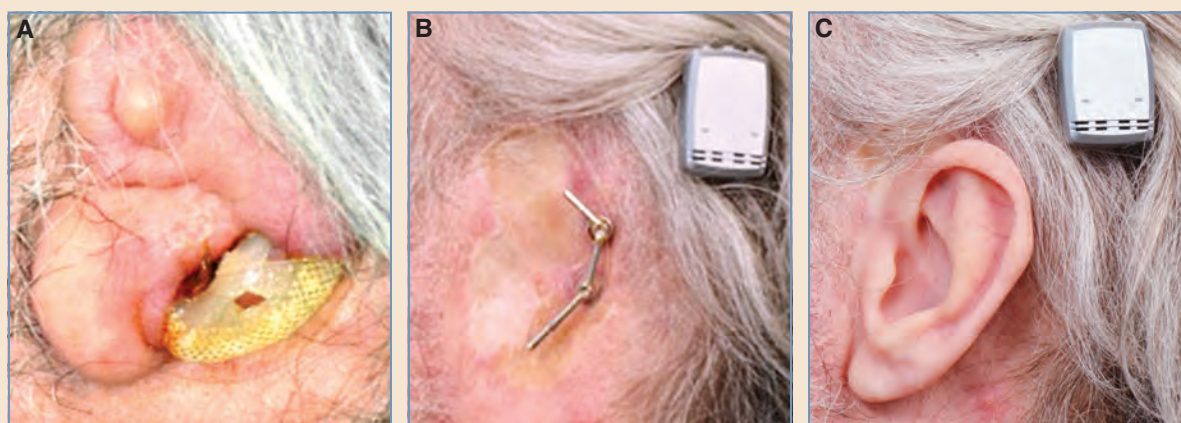


Fig. 30-31 Extrusion of an alloplastic implant of a previously reconstructed ear can be corrected with surgical placement of implants to retain a prosthetic ear that is custom designed and color matched to the patient's skin. **A**, This patient had an extruding implant. **B**, Osteointegrated screws were placed with a bar-and-clip retention device. **C**, An auricular prosthesis is attached.

Table 30-7 Comparison of Autologous Versus Prosthetic Ear Reconstruction

Autologous Reconstruction	Prosthetic Reconstruction
Two surgical procedures	One or two surgical procedures
Technique-sensitive surgery	Less complex surgery
Longer initial healing time	Daily maintenance care of abutments
Lifetime maintenance free	Lifetime care of prosthesis
Uses a patient's own tissue	Lifetime maintenance and patient follow-up
Natural skin color match	Control of sculptural form
Limited control of form with the patient's tissue	Aging of artificial materials

for each patient, and therefore generally require multiple, sometimes lengthy, appointments for the initial design, fabrication, and fitting. Contemporary, computer-based approaches such as three-dimensional imaging, three-dimensional printing, and computerized color formulation have contributed to decreased treatment times, with improved accuracy, efficiency, and repeatability in the design, fabrication, and remake processes.⁷³ Given the relatively short lifespan of silicone prosthetic devices, access to a skilled practitioner who can reliably provide replacement prostheses over the span of a patient's lifetime should be considered.⁷⁴

The surgical technique for prosthetic rehabilitation involves implantation through an incision posterior to the external auditory meatus. Careful preoperative planning is essential to ensure appropriate placement of implants for the planned auricular restoration. Computer-based virtual surgical planning tools have become a trademark technique for many clinical teams who regularly perform these procedures and have access to the appropriate technologies. This becomes particularly beneficial when the availability, quantity, quality, and morphology of bone are unreli-

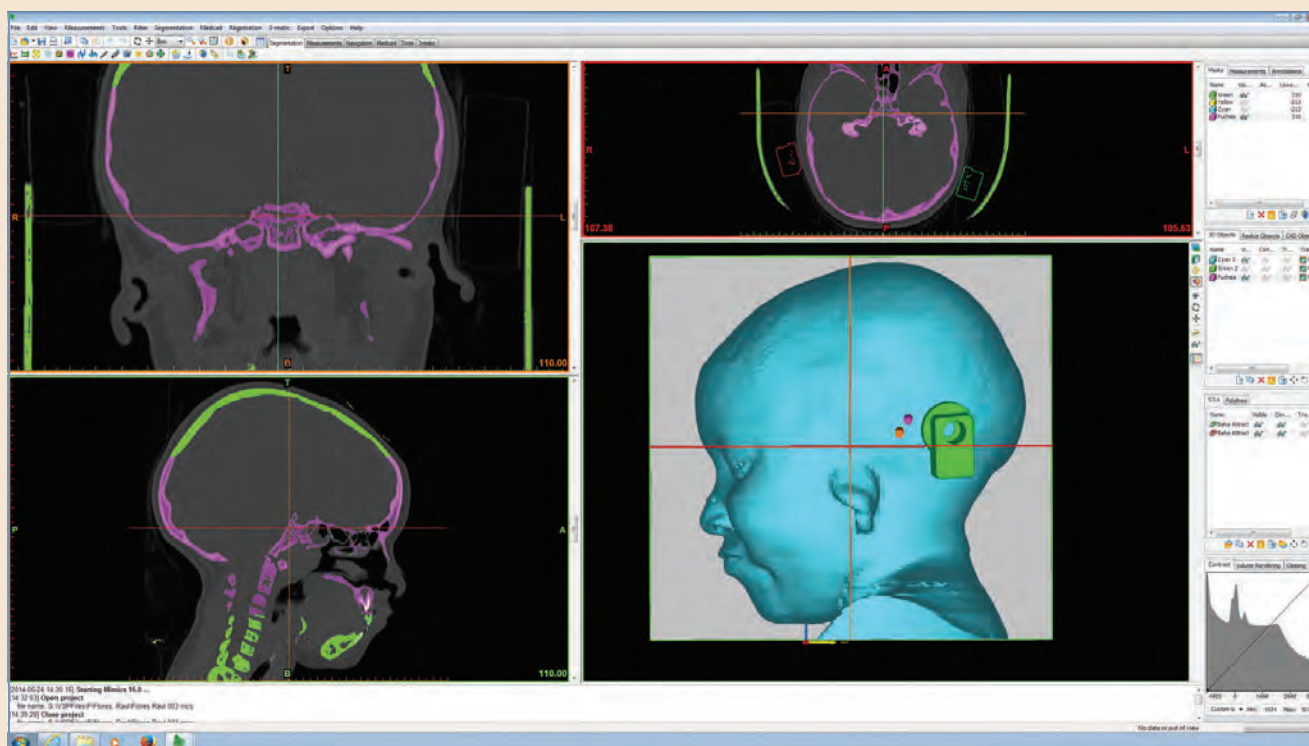


Fig. 30-32 Computer-based virtual surgical planning tools become essential when skeletal anatomy may be anomalous, particularly in CFM patients.

able, particularly in CFM patients (Fig. 30-32). Generally, in children, a two-staged procedure is preferred. In the first surgery, implants are placed in the bone and covered, and the skin is closed to allow osteointegration to occur. Three to four months later, the implant is uncovered and the percutaneous abutment is attached. After the skin heals (approximately 4 to 6 weeks), fabrication of the prosthesis begins. Healing times, based on the surgeon's assessment of bone quality during surgery, may vary with each patient. With abutment connection, revision of the subcutaneous tissue is required to achieve flat, tight adhesion of the skin to the bone during healing. This is essential to minimize adverse skin reactions of the periaabutment tissue and to facilitate the patient's ability to perform regular home hygiene to maintain tissue health.

Classifications have been defined to manage skin response of the periaabutment tissue.⁷² Auricular remnants, as in cases of microtia or failed reconstruction, can be excised at either the first or second surgery based on the surgeon's preference. Implant success is highest in uncompromised temporal bone. More implant failures are reported in the orbital bone and maxilla, sites common for retention of nasal and orbital prostheses. Implant failure is higher in irradiated bone. Where implant failure is a concern, additional implants are often placed as "sleeper" implants.⁷⁵ When implants are placed to retain an ear prosthesis and bone conduction device in the same surgical procedure, careful planning with an anaplastologist and audiologist is essential to ensure appropriate placement and adequate distance between implants.

Management of Hearing Loss in Patients With Craniofacial Microtia

Restoration of binaural hearing in patients with unilateral atresia has been shown to result in improved sound localization, distance hearing, and hearing in noisy environments and may be accomplished in a variety of ways.⁷⁶ Bone conduction hearing devices convert sound to vibration and are held against the scalp over the mastoid by a headband or metal spring clip. The disadvantages of these devices include high visibility and problems with skin irritation and breakdown from pressure.

Semiimplantable bone conduction devices (BCDs) are available. These can be direct-drive devices that transmit sound vibrations directly to the mastoid through an implanted percutaneous interface (Baha, Cochlear) or skin-drive devices, which are retained by an implanted magnet and transmit sound vibrations across intact skin. Direct-drive devices provide the most effective sound amplification; however, the percutaneous abutment requires daily care and may lead to problems such as skin irritation, overgrowth, and infection. Improved sound processing in the latest generation of skin-drive devices such as the Baha 4 Attract (Cochlear Bone Anchored Solutions) has made these a viable option for many patients.^{77,78}

Other options include transcutaneous direct-drive BCDs, such as the Bonebridge (MED-EL), which uses an implanted sound transducer and transmits only an electromagnetic signal across the skin. Preliminary results have been promising for this system.⁷⁷ Alternatively, a prosthesis (Vibrant SoundBridge, MED-EL) may be implanted in the middle ear, attached to either the stapes or oval window to stimulate only the ipsilateral ear and to potentially improve sound localization.⁷⁹ Canalplasty, or reconstruction of the external auditory meatus, canal, and tympanic membrane, can be attempted; however, this procedure requires a functioning middle ear and is associated with high complication and reoperation rates, and approximately 30% of patients will require a hearing aid postoperatively.⁷⁶

Macrostomia Reconstruction

Macrostomia is typically reconstructed in infancy to restore lip competence for oral feeding. The distance from midline to the unaffected oral commissure is used as a reference for the location of the reconstructed commissure. In bilateral cases, the demarcation between normal tissue and hypoplastic cleft tissue may be used instead. The commissure is reconstructed with triangular mucosal flaps, approximation of the oral and buccal musculature, and skin closure with a W-plasty.⁸⁰

Epibulbar Dermoid Cysts and Lipodermoids

The indications for removal of epibulbar dermoids and conjunctival lipodermoids are similar. Although these lesions are benign, they may pose a risk for visual obstruction with resultant amblyopia. Smaller cysts can be relatively inconspicuous; however, larger lesions may produce visible distortion of the iris and surrounding tissues, leading to obvious deformity. Children with epibulbar dermoids and lipodermoids should be referred to an experienced ophthalmologist for possible excision, which in many cases can be accomplished with minimal corneal scarring.⁸¹

Velopharyngeal Insufficiency

Palatal muscle hypoplasia may be present and can lead to velopharyngeal insufficiency in patients with CFM. In a series of 18 patients, Luce et al²⁰ reported significant velopharyngeal insufficiency in one third. The affected patients tended to be those with more severe hypoplasia of the maxillary-malar complex. Nasendoscopy and periodic speech evaluation should be used to

guide management, and surgical options are similar to those in the non-CFM population. However, the risk of obstructive sleep apnea in the presence of micrognathia should be considered.²⁰

Facial Nerve

Managing facial nerve palsy in patients with CFM is dependent on the pattern and severity of dysfunction, as with non-CFM patients. A complete discussion of facial nerve palsy and its management is provided in Chapter 38. However, in patients with hemifacial hypoplasia and concomitant facial nerve palsy, a free neurotized muscle-flap transfer, such as the gracilis or latissimus dorsi, can serve two purposes: restoration of dynamic facial animation and augmentation of tissue volume on the affected side.^{82,83} These techniques may be carried out in a single stage or as a two-stage procedure in combination with cross-facial nerve grafting and is an excellent option in these patients.

For patients with marginal mandibular nerve weakness, asymmetrical lower lip depressor muscle function can be mitigated by injection of botulinum toxin or by a surgical myectomy. The benefits of botulinum toxin injection are its convenience and minimally invasive nature; however, its effects are temporary, and repeated injections are required. Conversely, a myectomy can be a more permanent solution, but, even after this treatment, a small proportion of patients may require subsequent botulinum toxin injections for optimal efficacy.⁸⁴ Both techniques can be effective, and the choice should be tailored to the patient.

CONCLUSION

Restoring facial symmetry is one of the most challenging of all reconstructive procedures. Its difficulty lies in the extent of the involvement, ranging from purely skeletal asymmetry to involvement of all components of the facial structures. Restoring facial symmetry requires establishing an axis of symmetry, which becomes increasingly difficult with increasing severity of involvement. Although the craniofacial skeleton can be disassembled from the sphenoid and sectioned into its anatomic units, the challenge is in knowing where to position each of its bony elements relative to the facial axis and subsequently addressing the soft tissue surface and volumetric asymmetry. Today, surgeons, orthodontists, and anaplastologists have the benefit of viewing the three-dimensional asymmetry at multiple levels with cone-beam CT and three-dimensional surface cameras. These allow more accurate visualization of the morphologic deformity and planning of the surgical approach.

No other type of patient requires the same degree of imagination, innovation, and patience on the part of the surgeon and treatment team. Nearly every anatomic component can be affected by CFM. Similarly, challenges for patients and their families are equally difficult. From birth to early adulthood, and sometimes later, a child's habilitation can require numerous procedures, and a significant financial and time commitment on the part of the family. Many patients and their parents become tired, frustrated, and at times, uncooperative. Treatment should be planned with definitive goals in mind and conducted within a definite time frame so that these young patients do not think that their treatment is going on forever, with no end in sight. The orthodontist must accomplish as much as possible within a year in a first phase of treatment, not allowing a first phase to "melt" unnoticed into a second phase and subsequent orthodontic phases. Patients will notice, and the result may be a precipitous drop in rapport and cooperation as they become more frustrated and disheartened. Similarly, surgeons must have definite goals and carefully determine the number and timing of surgical interventions. Ultimately, success requires close working relationships between not only the surgeon and orthodontist, but also among the other members of

a supportive, multidisciplinary team, including psychologists, speech and language pathologists, prosthodontists, and anaplastologists. Paramount to any technically perfect, reconstructed face is the careful management of the patient's, parents', and family's expectations of treatment outcomes and the treatment experience itself. The most perfectly planned and executed treatment and symmetrically reconstructed face can be easily "dismantled" by unrealistic expectations of treatment. Honest, comprehensive, and compassionate education sets the foundation for successful treatment outcomes and a patient's physical, functional, and psychosocial rehabilitation.

KEY POINTS

- The spectrum of skeletal and soft tissue deformities seen in CFM arises when the normal developmental processes in the first and second branchial arches are disrupted.
- Many view microtia as a microform of the syndrome. Meticulous inspection will continue to reveal minor skeletal and soft tissue deformities of other branchial arch derivatives.
- Many classifications have been proposed based on either a single aspect of CFM (for example, the mandible, as in Pruzansky's classification) or the full spectrum of affected tissues (for example, OMENS-Plus). Some may prove more useful than others in treatment planning. The choice of classification system should be contextually based.
- Radiologic studies are necessary to fully define the anatomic deformity. CT with three-dimensional reconstruction offers refinement in anatomic and morphologic mapping to aid in the diagnosis, treatment planning, and post-surgical management.
- For comprehensive evaluation and management, these children should be seen regularly by a comprehensive craniofacial team.
- Airway and feeding are the two primary issues prompting early intervention. The role of distraction osteogenesis of the mandible in early decannulation and/or prevention of a tracheostomy is well documented.
- Upper airway compromise resulting from severe mandibular micrognathia in bilateral HFM may require consideration of bilateral mandibular distraction or the need for a tracheostomy.
- Macrostomia, because of an absence of the normal checkrein effect of the orbicularis muscle on oral competence, may result in failure to thrive. Early repair can significantly improve feeding.
- Children who are born with complete absence of the ramus, condyle, and temporomandibular joint fossa need reconstruction early in life.

- Delaying microtia reconstruction until patients are 10 years of age or older may allow better positioning of the reconstruction, particularly in cases of more severe deformity, when prior distraction or other skeletal reconstruction may have at least partially improved the symmetry of the underlying facial skeleton.
- In general, the success of soft tissue reconstruction depends on prior correction of the skeletal deformity. However, the treatment plan for each patient must be individualized, and patients with severe deformities may require soft tissue interventions well before the completion of skeletal growth.
- Modern imaging techniques, three-dimensional software reconstruction and Virtual Surgical Planning have eliminated much of the accumulated error associated with two-dimensional tracings and traditional model surgery, allowing more precision in complex movements of the mandible and maxilla.
- Patients and parents must understand that mirror-image symmetry cannot be achieved. Muscles that are not present cannot be created. A degree of asymmetry should always be expected.

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31

Microtia

Gordon H. Wilkes • Regan Guilfoyle



Reconstruction of microtia has always been a challenge. Early attempts were largely unsuccessful. A myriad of techniques and materials have been used with limited long-term success. Radford Tanzer's landmark publications in 1959¹ started a new era in microtia reconstruction. Burton Brent's contributions starting in the 1980s²⁻⁵ resulted in a significant improvement in the treatment possibilities for microtia patients and set a new standard for surgical care in microtia. Further refinements in technique by others starting in the 1990s, including Nagata,^{1,6-8} Firmin et al,^{9,10} and Park et al,^{11,12} have resulted in continual improvement in surgical results. Treatment selection in microtia is still not without controversy. The lack of surgical expertise to consistently achieve good results with these new autologous techniques and the variability of surgical results have led to the development of other treatment modalities. These will be discussed further in this chapter. Several excellent reviews are available for those interested in this field.¹³⁻¹⁹

MICROTIA CLASSIFICATION AND CHARACTERIZATION

Historically, many classification systems of major ear deformities have been described, including those by Marx (1926),²⁰ Rogers (1974),²¹ and Tanzer (1977).²² Most systems describe congenital deformities and classify them according to embryonic development, from mildest to most severe deformity. These are not particularly helpful for specific surgical planning.

Weerda²³ expanded on Marx and Roger's system by trying to include all dysplasias in three categories. More recently, Hunter et al²⁴ published a paper to standardize the terminology used to describe and classify ear dysmorphology based on Weerda's system (Table 31-1). Nagata⁶ classified microtia into five types (Fig. 31-1):

1. Lobule
2. Concha
3. Small concha
4. Anotia
5. Atypical microtia

These modified classifications have some relevance for definitive surgical planning; the more descriptive the name of the ear deformity, the more helpful it is in surgical planning.

Human Disorders Associated With Microtia

Patients with microtia should be examined for other dysmorphic features. It has been reported that 20% to 60% of children with microtia have an identifiable syndrome or other anomalies (Box 31-1). These include craniofacial microsomia, Townes-Brocks syndrome, and mandibulofacial dysostosis (Treacher Collins and Nager syndromes).²⁵ Oculoauriculovertebral spectrum (OAVS) includes facial asymmetry, ear and facial tags, epibulbar dermoid benign tumors, microphthalmia, macrostomia, and microtia (Fig. 31-2). Extracranial features include renal, cardiac, and vertebral anomalies. Goldenhar syndrome and hemifacial microsomia are included in this spectrum. Most cases are sporadic with occasional genetic inheritance. There remains controversy whether isolated microtia is a mild phenotype of OAVS. The present thinking is that microtia and OAVS should be considered separate entities.

Table 31-1 Hunter Classification of Microtia: Modification of the Weerda Classification

Degree	Definition
First	Presence of all the normal ear components and the median longitudinal length more than 2 standard deviations below the mean
Second	Median longitudinal length of the ear more than 2 standard deviations below the mean in the presence of some, but not all, parts of the normal ear Replaces: Grade III dysplasia, severe cupped ear, cockleshell ear, type IV constricted ear, snail ear, shell ear, mini ear
Third	Presence of some auricular structures, but none of these structures conform to recognized ear components Replaces: Grade III dysplasia, group III hypoplastic ear, peanut ear

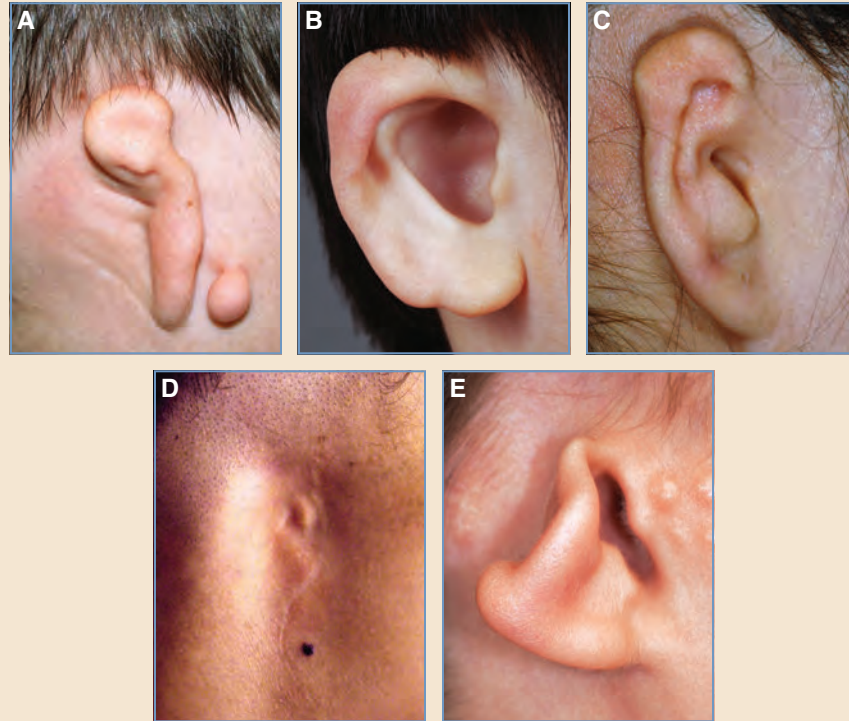


Fig. 31-1 Microtia types. **A**, Lobule. **B**, Conchal. **C**, Small conchal. **D**, Anotia. **E**, Atypical.

Box 31-1 Microtia-Related Anomalies

Oculoauriculovertebral spectrum disorders

Facial asymmetry
Ear and facial tags
Epibulbar dermoids
Microphthalmia
Macrostomia
Microtia

Extracranial anomalies

Renal, cardiac, and vertebral anomalies
Goldenhar syndrome
Hemifacial microsomia

Other

Craniofacial microsomia
Townes-Brocks syndrome
Mandibulofacial dysostosis
Treacher Collins syndrome
Nagar syndrome



Fig. 31-2 OAVS patient with microtia.

Epidemiology and Genetics

The reported prevalence of microtia varies from 0.83 to 17.4 per 10,000 births.²⁵ The etiologic factors are not clearly understood. There are two prevailing theories; the most current hypotheses for microtia include (1) neural crest cell disturbance, and (2) vascular disruption through several different mechanisms. There is evidence for a significant genetic and environmental contribution to microtia. The factors quoted include race, with a higher incidence in Asian, Hispanic, and First Nation populations, and drug usage, with a higher incidence associated with retinoic acid, alcohol, thalidomide, and mycophenolate mofetil, an anticancer medication. Microtia appears more frequently in mothers who were exposed to higher altitudes during pregnancy and those with type 1 diabetes. Mendelian inheritance is more common in syndromic and familial cases. Multifactorial or polygenic causes are more probable in sporadic cases. Microtia affects the right ear 60% of the time and is 20% to 40% more common in males.

Embryology

The middle and external parts of the ear are derived from the first (mandibular) and second (hyoid) branchial arches.²⁶ The ear is formed from six hillocks that ultimately grow and coalesce along these arches. They are first seen at 5 weeks' gestation. During the course of development, the ear forms, grows, and migrates from an antero-caudal to a posterocranial position. Ultimately, 85% of the fully formed ear is derived from the hyoid arch. Only the tragus and crus helix are derived from the mandibular arch. The canal and concha are derived from the first branchial cleft. The inner ear, first appearing at 3 weeks' gestation, has an ectodermal embryologic origin, so it is not typically involved in microtia.

PREOPERATIVE PLANNING

Initial Consultation

Early consultation helps the surgeon to develop a trusting relationship with the family. Because of their easy access to the Internet, parents may be bombarded with information that may be factual and informative, biased, or frankly deceptive. The surgeon can guide them through a discussion

of ear reconstruction and treatment options, managing expectations and at all times striving to alleviate parents' feelings of guilt.

Psychosocial Issues

The prevalence of mood disorders is high in microtia patients, with “depression” in 20.2%, “interpersonal sensitivity/social difficulties” in 36.6%, and “hostility/aggression” in 26.3%.²⁷⁻²⁹ These percentages were higher in older patients who had not undergone reconstructive surgery. The age at surgical intervention must be balanced against the potential for a good aesthetic reconstruction; a poor result may have a long-term adverse psychological effect on the individual's self-esteem. The child usually discovers he or she is different at about age 3 or 4. The earlier children are made aware that their ear is different, the lower the prevalence of psychological disorders. This is one factor to consider when discussing the timing of reconstruction. Both Brent⁵ and Jiamei et al³⁰ felt the major determinant of these psychological issues is transmission of parental guilt. Teasing, severity of microtia, low levels of maternal education, family disharmony, parental anxiety, and overprotection were all significantly associated with mood disorders in microtia patients.

Treatment Selection

The general principle of reconstructive surgery to use autologous tissue whenever possible also applies to ear reconstruction. The most common treatment choice for microtia is autologous reconstruction using costal cartilage. The surgical techniques required to obtain consistent results are very challenging and difficult to learn. This has led to a wide variety of surgical outcomes. Because of this, two alternative methods for microtia reconstruction have developed and have strong proponents. These are:

1. The use of a porous polyethylene ear framework covered by a temporoparietal fascial flap and skin graft
2. Removal of the microtic remnant, placement of osteointegrated implants, and use of a prosthesis

SURGICAL OPTIONS FOR AUTOLOGOUS EAR RECONSTRUCTION

The choice of procedure is based on the surgeon's experience and training. A reasonable result should be obtainable with any of these procedures when done correctly.

The Brent technique^{2,3,5} set the standard for surgical care of microtia starting in the 1980s. His technique consists of four stages:

1. Rib harvest and framework creation
2. Lobule transposition
3. Framework elevation
4. Tragus reconstruction, conchal excavation, and contralateral otoplasty, if needed

Further evolution in technique led by Nagata,^{7,8,31} Firmin,⁹ and others^{12,32,33} has led to fewer surgical stages with a more complicated three-dimensional (3D) framework design. They have also changed the approach to soft tissue management of microtia. Their first stage combines framework creation, lobule transposition, and tragal reconstruction. The second stage is elevation of the framework, placement of a cartilage block under the framework to help maintain projection, and coverage with a temporoparietal flap and a split-thickness skin graft from the scalp. Proponents of these newer techniques cite improved contour of the reconstructed ear, including better definition of the tragus, antitragus, antitragal notch, and antihelical fold. They also think there

Table 31-2 Comparison of Autologous Ear Reconstruction Techniques

	Age at Harvest	Side of Harvest	Framework Construction	Plane of Rib Dissection	Fixation Technique	Postoperative Dressings	Number of Stages
Brent ⁵	5 or 6	Contralateral	Two piece	Supraperichondrial	Nylon suture	Suction drains	4
Nagata ⁶	10 (or when the circumference at the xiphoid process is 60 cm)	Ipsilateral	Five piece	Subperichondrial	Stainless steel wires	Bolsters	2
Firmin ⁹	10	Ipsilateral	Five piece	Subperichondrial	Stainless steel wires	Suction drains	2

is better projection using cartilage during the framework elevation stage. Critics argue that the complication rate is higher, the techniques more difficult to learn, and the potential for chest wall deformity is higher. These three approaches are summarized in Table 31-2. Other autologous techniques and modifications used are based on the techniques of Brent,⁵ Nagata,⁶ and Firmin.⁹

Templating

Preoperative planning requires creation of intraoperative templates for positioning the new ear as well as creating the new 3D framework. The anatomic position of the reconstructed ear is a constant measure shared by all techniques. Proper placement of the new ear is a key step in creating a natural appearance. The ear should be placed according to appropriate anatomic landmarks, regardless of the position of the hairline or an existing remnant. If a compromise is to be made, it is important to note that an abnormal inferior-anterior position is more noticeable, so movements in these directions should be limited. Absolute measurements may need to be compromised in patients with facial asymmetry so that the ear fits in the relative correct position based on facial features.

In unilateral microtia, ear positioning is guided by the normal contralateral side. An x-ray film or other transparent celluloid material is placed on the nonmicrotic side of the patient's face and the ala, lateral canthus, eyebrow, lateral commissure, and normal ear are traced. This positioning template can then be superimposed on the microtic side of the face to reveal the correct relationship of these landmarks to the site of the future reconstructed ear (Fig. 31-3).

An ear template can be created by tracing the outline of the normal contralateral ear or a 3D model of the ear on a transparent film paper and then drawing the underlying "ear skeleton," removing 1.5 to 2 mm to account for the future thickness of the mastoid skin coverage (Fig. 31-4). Nagata has created a series of templates based on the normal anatomic relationships and dimensions of the ear (S. Nagata, personal communication, 2015). These templates come in varying sizes from 55 to 70 mm (Fig. 31-5). He chooses one that is 2 mm shorter than the normal opposite ear. He does not use the contralateral normal ear as a template.

Advanced digital technologies can provide the surgeon with 3D models of the "new" reconstructed ear using a moulage, CT scan, or laser scan of the normal ear and then reversing it. In patients with bilateral microtia, placement of the ear becomes a more difficult task. Templates

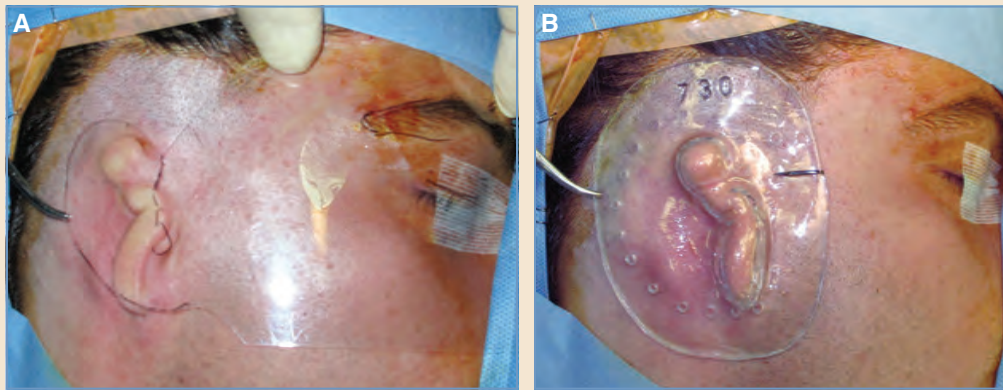


Fig. 31-3 Ear positioning. **A**, Using a clear template. **B**, Using an acrylic template.

Fig. 31-4 Two-dimensional transparent film template and 3D model.

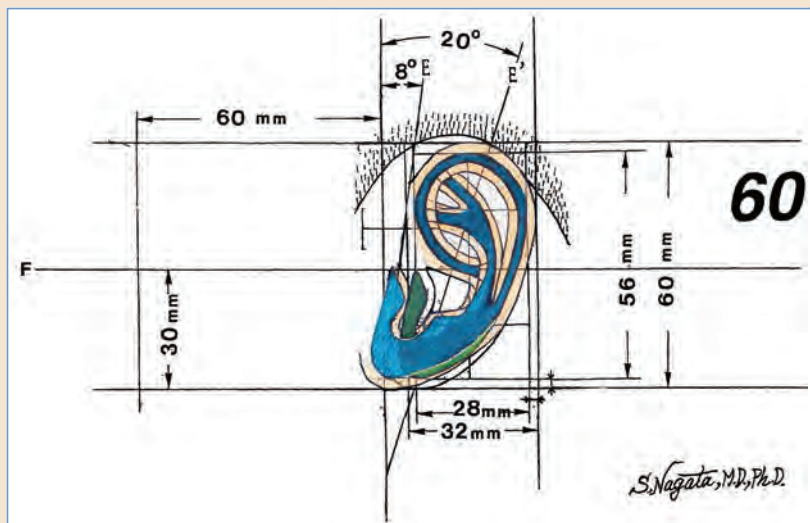


Fig. 31-5 Classic Nagata 60 mm template. (Courtesy of S. Nagata, MD, PhD.)

must be created so that consistency can be achieved on both sides. The actual ear template can be chosen from other patients or made from a family member's ear. A smaller ear (55 to 60 mm) is usually chosen, because less cartilage will be needed and less potential stress will be put on the skin envelope.

Rib Harvest Techniques

Rib is harvested with the patient in the supine position; harvesting can be done in the same procedure with preparation of the auricular pocket.³⁴⁻³⁶ The resultant chest wall deformity after rib harvest is a concern for critics of autologous reconstruction. Several authors have described a strategy to try to mitigate this concern.³⁷⁻³⁹ The incision is generally placed in a transverse or oblique orientation on the chest wall. The exact site and length of the incision can vary, depending on which ribs are harvested and the number of ribs required. Generally, the incision is made over the seventh rib and is a minimum of 5 cm in length (Fig. 31-6). In older females with mature breast development, the incision can sometimes be hidden in the inframammary fold. During rib harvest it is preferable to leave the rectus abdominis and external oblique muscles intact if possible, because transection of these muscles increases postoperative pain. Cautery should be used very judiciously on the perichondrium, since heat is damaging to chondrocytes and will cause later cartilage resorption.

The Brent technique⁵ uses a lower profile two-piece framework that requires less costochondral cartilage than other autologous techniques and thus can be performed in patients at a younger age, typically at age 5 or 6. Generally, the contralateral sixth, seventh, and eighth ribs are harvested. Dissection is performed in an extraperichondrial plane. Brent leaves a superior rim of the sixth rib to minimize chest wall deformity. In our experience, the incidence of an inadvertent pleural tear is higher with an extraperichondrial harvest than a subperichondrial harvest.

Creating the larger Nagata 3D framework⁶ requires more cartilage. He recommends delaying reconstruction until the patient has a total chest circumference of 60 cm measured at the xiphoid process, usually at about age 10. We have performed this technique on patients as young as 8 years of age if their chest circumference was adequate. The ipsilateral sixth, seventh, eighth,



Fig. 31-6 Landmarking the site for chest incision.

and occasionally ninth ribs are harvested in a subperichondrial fashion. Specialized instruments are available to elevate the perichondrium, which minimizes damage to the underlying cartilage (Fig. 31-7). The perichondrial sleeve is carefully incised down its midline but is otherwise left in situ. The only perichondrium excised is that which immediately overlies the sixth and seventh synchondroses.

During harvest, utmost care should be taken when handling the rib cartilage. The ribs are quite strong when traction is applied in a longitudinal direction, but flexing the rib can result in breakage. This is most likely to occur if the rib has been inadvertently nicked during dissection or has small areas of calcification or spongy deterioration within it (Fig. 31-8).

To try to minimize chest wall deformity, any cartilage pieces left over after the framework has been reconstructed are then diced into small pieces and either reintroduced into the perichondrial sleeve, or more recently, surgeons have placed them in a Vicryl mesh sleeve, making a



Fig. 31-7 Subperichondrial rib harvest with gouge.



Fig. 31-8 Harvested ribs.



Fig. 31-9 Pseudorib with Vicryl mesh sleeve.

pseudorib (Fig. 31-9). There is some evidence to suggest that this decreases the resultant chest wall deformity. Some surgeons, before skin closure, will bank a crescent-shaped wedge of cartilage subcutaneously in the incision site for future harvest during the second stage of reconstruction. Kawanabe and Nagata³⁸ often later use the cartilage regenerated within the pseudorib for framework elevation. Firmin's approach to harvesting^{9,10} is quite similar to Nagata's.

During rib harvesting the surgeon must be careful to avoid entering the pleural space. If the parietal pleura has been injured, it can be easily managed by inflating the lung, suctioning out the air, and closing the pleural defect. A chest tube is not needed unless the visceral pleura has been injured. Pain at the donor site can be mitigated with either an intercostal block or an indwelling local anesthetic catheter during the postoperative period. Drains are not usually necessary and add to the patient's chest discomfort.

Ear Framework Creation

Carving rib cartilage is best performed at a separate table using a combination of scalpel and gouges (Fig. 31-10, *A* and *B*). The finished framework should be devoid of sharp edges and have the appearance of a single, unified, sculpted piece. The pieces of the framework are fastened together using either suture material (Brent⁵) or double-armed stainless steel wire (Nagata⁶ and Firmin⁹) (Fig. 31-10, *C*). Regardless of the fixation used, the ends of the fixation material must not be exposed on the anterior surface of the framework, since this would increase the risk of extrusion.

Brent Framework

The Brent framework includes a base frame carved from the sixth and seventh ribs and a helical rim carved from the eighth rib.⁵ The contours of the antihelix are achieved by using a gouge to create the corresponding areas of relief on the base framework. No additional cartilage is added. The helical rim sits around the base frame and is sutured in place with horizontal mattress sutures of 4-0 clear nylon (Fig. 31-11).



Fig. 31-10 A, Instrument setup for carving rib cartilage. B, Specialized gouges. C, Stainless steel wires.

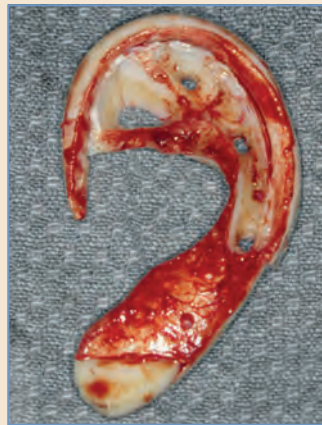


Fig. 31-11 Example of the Brent framework.

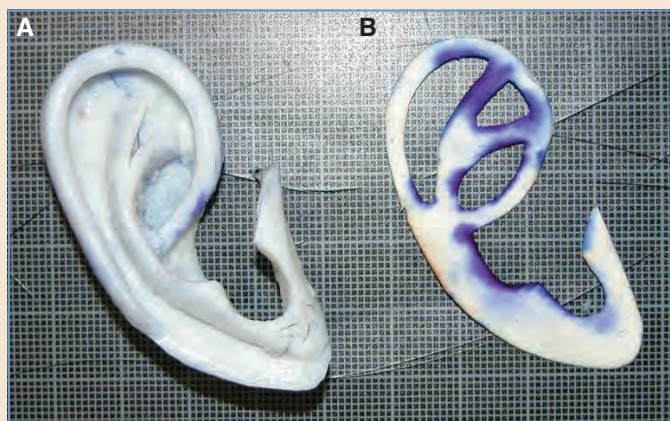


Fig. 31-12 Nagata framework. **A**, Cartilage frame. **B**, Paper template. (Courtesy of S. Nagata, MD, PhD.)

In Brent's classic description,² the tragus was not part of the initial framework. It was instead created from a composite graft from the contralateral conchal bowl during the fourth and final stage of reconstruction. Brent in some cases will add a tragal strut to his framework, which is particularly useful in cases of bilateral microtia.

Nagata Framework

Nagata uses a five-piece framework constructed from the sixth to ninth ribs.⁷ This framework includes a base frame, a helical rim, an antihelix, a tragal extension, and a conchal bowl graft for added projection (Fig. 31-12). In cases in which the skin pocket is tight or added projection is not needed, the conchal bowl unit can be omitted.

Pieces are affixed to the base frame with double-armed straight needle sutures of 38-gauge stainless steel wire. The wires are twisted and cut on the posterior surface of the framework. The loop of wire on the external surface is countersunk. Further definition is achieved by using a gouge to deepen the regions of the scapha and fossa triangularis on the base frame. Nagata has demonstrated his method of framework construction in the accompanying video entitled Ear Reconstruction: Nagata Framework.

Firmin Framework

The Firmin framework is similar to Nagata's, except Firmin does not carve a conchal unit but rather includes a "surélévation" (elevating) piece.⁹ This piece spans the root of the helix and the tragus and is attached to the posterior surface of these structures. The purpose is to give more projection and stability to the tragus and the root of the helix, because the contractive forces on the framework often cause blunting of these features. Firmin demonstrates her method of framework construction in the accompanying video entitled Ear Reconstruction: Three-Dimensional Costal Cartilage Frame.

Incisions and Soft Tissue Management

Correct surgical planning of incisions, meticulous dissection of the auricular pocket, and gentle handling of the surrounding soft tissues are critical components of successful ear reconstruction. Once the surgeon has perfected the skill of carving an ear framework, soft tissue management becomes the major determinant of aesthetic success in reconstructing an ear.

The microtic cartilaginous remnant is of no value and is always excised during the creation of the auricular pocket. The type of incision used to create the auricular pocket varies, depending on the ascribed technique and the type of microtic deformity. Postoperative management of soft tissue in all techniques is largely focused on skin circulation and preventing seroma or hematoma formation. This complication will thicken and fibrose the skin flap, blunting auricular definition and downgrading the aesthetic result.

In the first stage of the Brent technique^{2,5} the lobule is left in situ, and a longitudinal skin incision is made posterior to the auricular vestige. A thin skin flap is elevated, taking care to preserve the subdermal vasculature and the microtic cartilaginous remnant is removed. The pocket is further developed to 1 cm outside the ear markings. The two-piece framework is introduced into this pocket. Skin is closed over two suction drains, one beneath the framework and the other adjacent to the helical rim. Epinephrine solutions are not used so that skin circulation can be evaluated during the procedure. Brent feels the use of skin bolsters increases the risk of skin necrosis (Fig. 31-13).

Nagata^{6,7} was the first to describe a skin incision technique that would allow rotation of the lobule and creation of an auricular pocket in a single stage. A W-shaped incision is placed along the border of the mastoid region, extending vertically on the posterior aspect of the lobule, crossing the lobule anteriorly, and ending in a small 2 mm circular incision that forms the U-shaped configuration of the intertragal notch. These incisions unfold to form three flaps, the anterior and posterior skin flaps of the tragus and the anterior skin flap of the lobule⁶ (Fig. 31-14).



Fig. 31-13 Brent framework with suction drains.

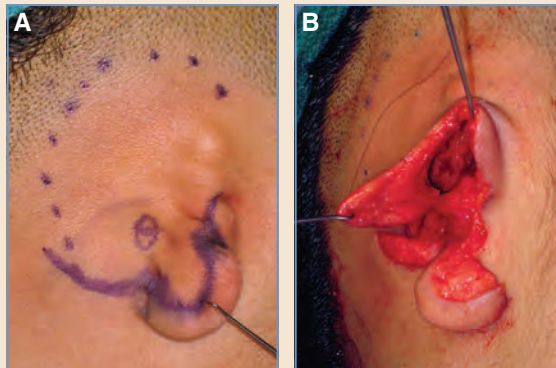


Fig. 31-14 Nagata W-shaped incision. **A**, The incision with a pedicle. **B**, The incision with remnant removed.

A small subcutaneous pedicle is left above the central limb of the W flap.⁶⁻⁸ The pedicle has been demonstrated to enhance the blood supply to the skin flaps.^{39a,39b} The remnant is then removed and a slightly oversized auricular pocket is dissected. Once the skin flaps and auricular pocket are created, the tragal portion of the framework is tucked around the subcutaneous pedicle, and the superior portion of the framework is rotated into the remainder of the pocket. If the addition of a tragal strut or “surélévation” piece, as described by Firmin,⁹ is desired, this piece must be fixed after the placement of the framework in the pocket.

The W incision can be modified for cases of small and large conchal-type microtia. The skin incision for a small conchal-type microtia³¹ is virtually identical, except that the anterior lobular incision must be placed posterior to the indentation created by the small concha. The incision modifications required for the large conchal-type microtia are more dramatic.⁸ The tragal and lobular portions of the ear are intact and are thus not reconstructed in the 3D framework. The posterior skin incision has a similar W-shape but is placed more superiorly and is extended anteriorly across the helix to the antihelix, terminating on the posterior conchal wall.

Nagata⁶ uses temporary suction to hold the skin flaps in position against the framework and then places bolsters. The bolster dressing is checked every day, and antibiotic ointment is applied daily. Bolsters are kept in position for 10 to 14 days (Fig. 31-15). The flaps created bring more tissue transversely across the framework, so the skin is under less tension when the bolsters are placed. In our experience, if skin necrosis is to occur with the Nagata technique, it is most likely to occur on the posterior aspect of the tragus at the tip of the W flap. In the Brent technique, it is more likely to be over the helical rim.

Firmin has developed an algorithm for incision choice based on size and shape of the remnant.¹⁰ A type 1 incision for classic lobular-type microtia is like a Z-plasty with the lobule as one of the flaps. A type 2 incision for conchal-type microtia is transversely oriented at the midpoint of the remnant and includes a backcut to allow closure of the posterior incision to the mastoid skin. Firmin¹⁰ does not maintain a subcutaneous pedicle. Fig. 31-16 demonstrates the Brent, Nagata, and Firmin approaches for skin incisions of lobular-type microtia; the Nagata and Firmin approaches for small conchal-type microtia; and a universal approach for anotia.

Other Reconstructive Stages

The second stage in the Brent technique^{2,5} consists of rotating the lobule from a vertical to a horizontal position. The framework is elevated in the third stage. Initially Brent used a split-thickness skin graft to cover the posterior aspect of the framework after elevation, but because of the loss of the sulcus and projection, he changed his technique. An incision is made posterior to the framework and a banked piece of cartilage from the chest wall or behind the ear is retrieved. This wedge of cartilage is placed under the framework and is covered with a retroauricular fascial flap. A full-thickness skin graft is then harvested from the lower abdomen or groin and placed over the auricular sulcus defect.

The fourth and final stage is tragal reconstruction, conchal excavation, and, when necessary, reduction of the contralateral ear for improved symmetry. The tragus is constructed from a composite skin-cartilage graft from the contralateral ear and is inserted along a J-shaped incision placed at the posterior tragal margin. Some surgeons think that this often produces an unnatural appearance with an acute intertragal notch and poor definition of the conchal bowl (Fig. 31-17).



Fig. 31-15 Bolster dressing.

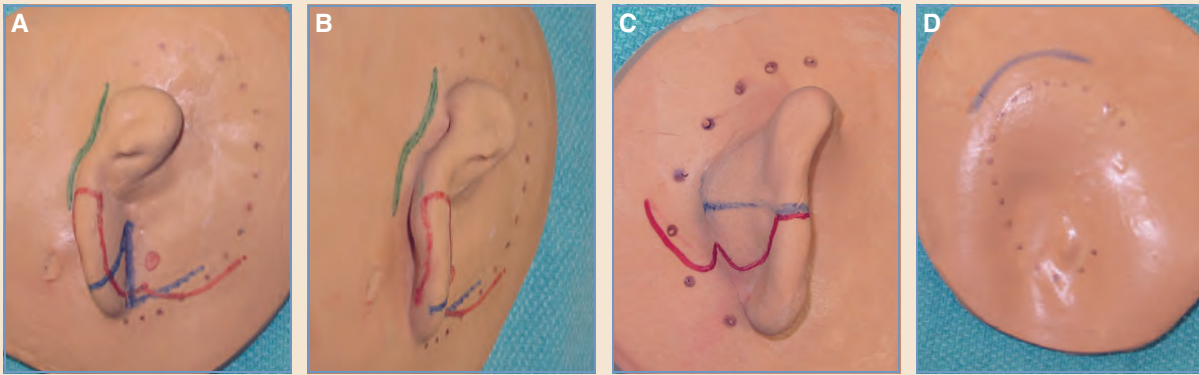


Fig. 31-16 Skin incisions for lobular-type microtia. Brent (*green*), Nagata (*red*), and Firmin (*blue*) approaches. **A**, Posterolateral view. **B**, Anterolateral view. **C**, Small conchal-type. **D**, Anotia.



Fig. 31-17 Tragus reconstructed with contralateral conchal bowl graft.

By adding a tragal unit to the 3D framework and creating a skin flap that naturally incorporates transposition of the lobule, Nagata was able to successfully combine the first three stages of the Brent technique into a single stage. Elevation of the framework is performed in the second stage, 6 to 12 months after the first stage. An incision is made 5 mm posterior and superior to the framework, and the framework is undermined to the level of the conchal bowl. The previously banked crescent-shaped rib graft is retrieved from the chest wall incision site, placed under the 3D construct, and fixed in place.

Next, a thin ultradelicate split-thickness skin graft (UDSTS) is harvested from the scalp using a No. 15 blade scalpel or dermatome. This graft is harvested well above the level of the hair follicles and produces a thin graft that is an acceptable color match. A temporoparietal fascia flap (TPF) is harvested through a zigzag or transverse incision and tunneled subcutaneously to cover the posterior surface of the projected auricle. The UDSTS is sutured in place over the TPF flap and further secured with a bolster dressing (Fig. 31-18).

The Firmin technique¹⁰ also involves two stages. Firmin advocates aggressive framework elevation. The banked wedge of cartilage is fixed to the undersurface of the framework at the level of the antihelix. The posterior skin is advanced and affixed to the depth of the sulcus. The exposed auricle is then covered with a TPF flap. A split-thickness skin graft is harvested from the scalp using a dermatome and is cut into two grafts. One graft is placed on the posterior aspect of the auricle and the other is placed on the mastoid surface.

Chen et al⁴⁰ describes a technique elevating a thin split-thickness skin graft from the hair-bearing scalp in continuity with the full-thickness skin posterior to the ear to cover the TPF, avoiding some visible scarring. A study comparing several techniques of sulcus construction showed no difference in ear elevation at 3 months.⁴¹

Regardless of the technique used, primary graft take is crucial to prevent secondary wound contraction and loss of projection. Because of the variability of results after ear elevation and deterioration of the aesthetic outcome, some surgeons have abandoned this second stage⁴² (Fig. 31-19).

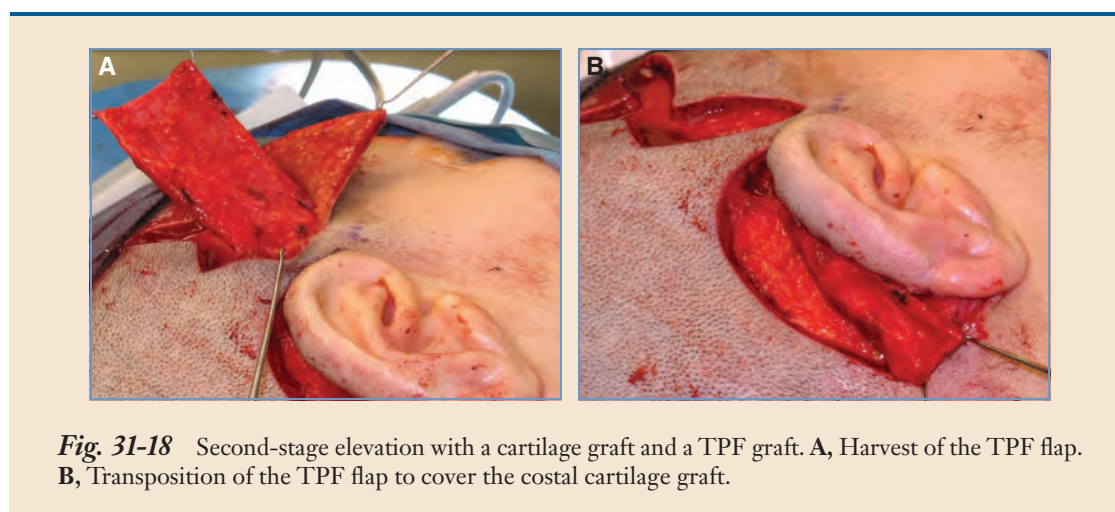


Fig. 31-18 Second-stage elevation with a cartilage graft and a TPF graft. **A**, Harvest of the TPF flap. **B**, Transposition of the TPF flap to cover the costal cartilage graft.



Fig. 31-19 Full reconstruction. This patient with lobular-type microtia is shown through the course of a two-stage reconstruction. A BAHA cochlear implant is placed at the time of the second stage. **A**, Lobular-type microtia vestige. **B**, Completed cartilage framework. **C**, The skin flaps elevated before placement of the framework. **D**, At closure, the skin flaps drape into the contours of the cartilage, aided by a suction drain. **E**, A bolster dressing further contours skin to cartilage. **F**, The BAHA guide, shown at the time of the second-stage elevation of the reconstructed ear. **G**, The temporoparietal fascia flap. **H**, The banked cartilage graft in place and covered with the temporoparietal fascia flap. **I**, The scalp donor site and the split-thickness skin graft in place over the postauricular fascial flap and remainder of the postauricular sulcus. **J**, The skin graft covered with a bolster dressing, and a dressing on the donor site at completion of the second stage. **K**, The completed reconstruction showing the sound processor in place. **L**, The BAHA abutment with the well-healed skin graft visible on the postauricular surface.

Patients With Low Hairlines

It is not uncommon to have a small amount of hair growth on the superior edge of the reconstructed ear. If the amount of hair growth is limited, this can be managed by trimming, laser therapy, or electrolysis. In cases of true low hairline, appropriate ear positioning is not compromised. Laser hair removal can be done before any surgical intervention is undertaken. Nagata describes a technique in which the auricular pocket is raised as an inferiorly-based flap with the hair-bearing portion raised as an UDSTS graft. The superior portion of the framework is then resurfaced with a TPF flap and covered with the attached UDSTS (Fig. 31-20).

Secondary Cases

Not all reconstructed ears are aesthetically acceptable. Poor results can occur from a surgeon's inexperience or complications such as hematoma, major infection, or skin necrosis. Patients are often reluctant to undergo a repeat costal cartilage graft harvest. Treatment of failed autologous ear reconstructions is a difficult challenge and speaks to the importance of achieving a satisfactory result the first time.

Autologous salvage procedures require either partial or complete reconstruction of the 3D framework. Rib harvest is performed on the opposite side of the original harvest to ensure a sufficient amount of cartilage is available. The skin flaps are often thick and fibrosed and offer limited coverage of the new framework. If the TPF flap is still intact, this flap can be raised and used to cover the framework, followed by a split-thickness skin graft.⁴³ If the TPF flap is unavailable, the deep temporoparietal fascia can be used, but it is less reliable. The chances of achieving a reasonable aesthetic result in this setting are much more challenging (Fig. 31-21). Soft tissue expanders can occasionally be of benefit to provide some coverage for expanding nonscarred skin only.⁴⁴ Expanding scarred skin or skin grafts has a high complication rate and cannot provide the quality of skin required in ear reconstruction.

Another option includes removal of the ear reconstruction, followed by placement of osteo-integrated implants and creation of an ear prosthesis (Fig. 31-22).

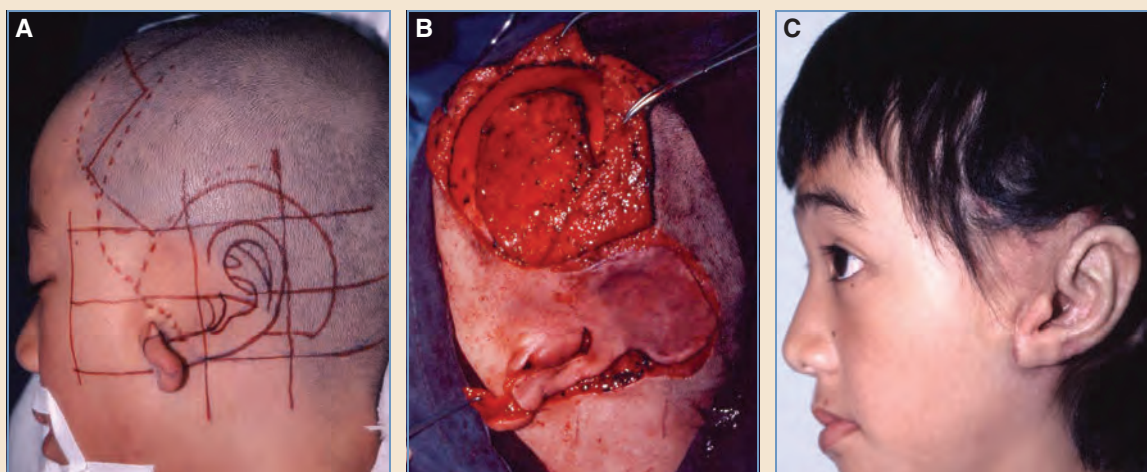


Fig. 31-20 Low hairline procedure. **A**, Preoperative markings. **B**, Intraoperative view with harvest of the UDSTS graft. **C**, Postoperative results. (Courtesy of S. Nagata, MD, PhD.)

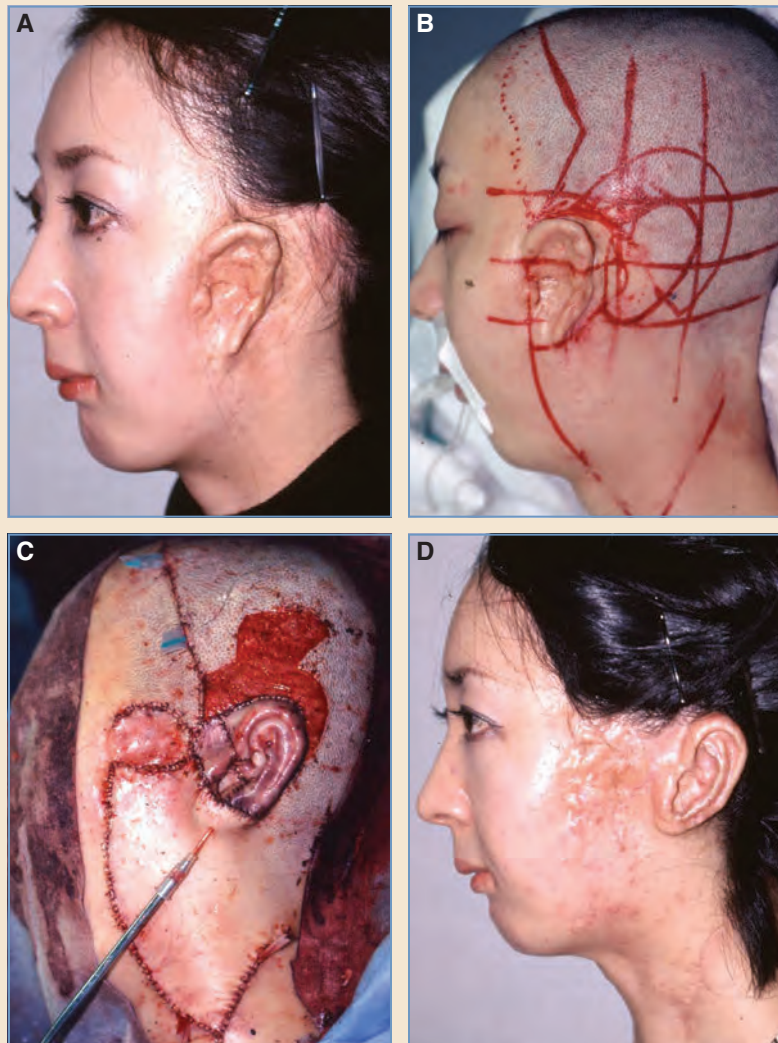


Fig. 31-21 Secondary autologous reconstruction. **A**, Preoperative presentation. **B**, Preoperative markings. **C**, Intraoperative dissection. **D**, Postoperative results. (Courtesy of S. Nagata, MD, PhD.)

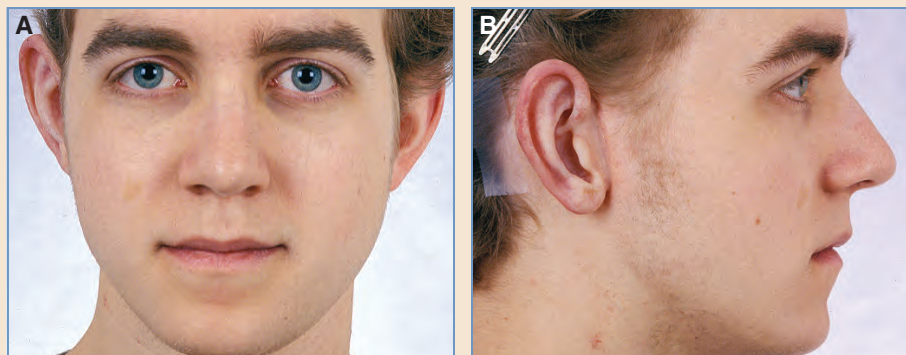


Fig. 31-22 Osteointegration with an ear prosthesis. **A**, Frontal view. **B**, Lateral view.

Other Reconstructive Options

Porous Polyethylene Frameworks

Because of concerns of chest wall morbidity, the challenge of carving an ear framework, and the variability of surgical results, alternative methods of ear reconstruction have been explored. Early alloplastic frameworks such as Silastic are no longer used because of the potential for long-term complications, including infection and extrusion. The use of porous polyethylene for the framework initially had similar problems. However, Reinisch, a pioneer in the use of porous polyethylene, has refined the technique to include complete coverage of the framework with a TPF flap in all cases.⁴⁵ The TPF flap improves the vascularity of coverage, decreasing the complication rate. It also allows some movement of the graft over the framework, which may have a protective effect. He has shown excellent aesthetic results obtained at an earlier age⁴⁶ (Fig. 31-23).

Reinisch⁴⁵ often harvests the large TPF flap using an endoscopic technique through a post-auricular incision. The TPF flap must be of sufficient size to cover the framework in its entirety. The auricular skin pocket is also dissected from this same incision. A splint is placed over the reconstruction initially to reduce the risk of seroma or hematoma. He has had success with both low hairline and secondary reconstructions.

The advantage of this approach is that it is a one-stage procedure performed on an outpatient basis with no rib harvesting. Critics argue that it invites less-experienced surgeons to attempt ear reconstruction, yielding poor results.⁹ It uses the valuable temporoparietal fascia, making it unavailable for any salvage procedures. They would argue that Reinisch's results are hard to duplicate, and late extrusion of the alloplastic framework is always a possibility. Long-term studies will help to further define the role of these treatment options in the management of microtia.

Osteointegrated Prosthetic Reconstruction

Osteointegrated implant-retained (OPR) prosthetics are a useful treatment option in adults with major ear deformities from a variety of different causes, but their use in children with microtia is controversial.^{46,47} The procedure requires removal of the ear remnant and creates scarring in the region of the ear, severely compromising future autologous options. Children may have an intermittent soft tissue reaction around the abutments and will need a new prosthesis every 2 to

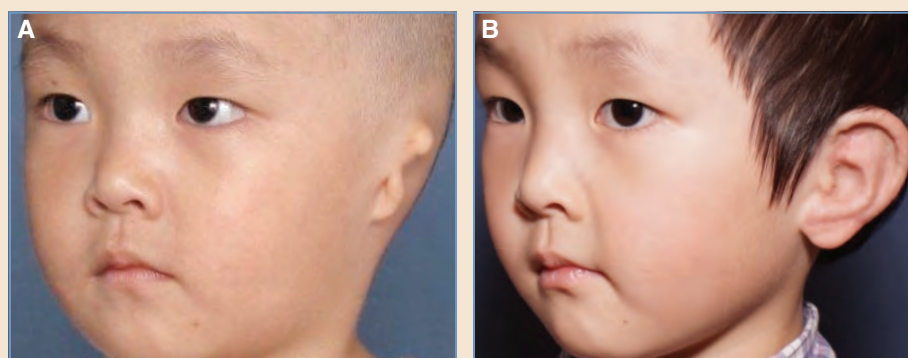


Fig. 31-23 Ear reconstruction with porous polyethylene. **A**, Preoperative view. **B**, Postoperative result. (Courtesy of J. Reinisch, MD.)

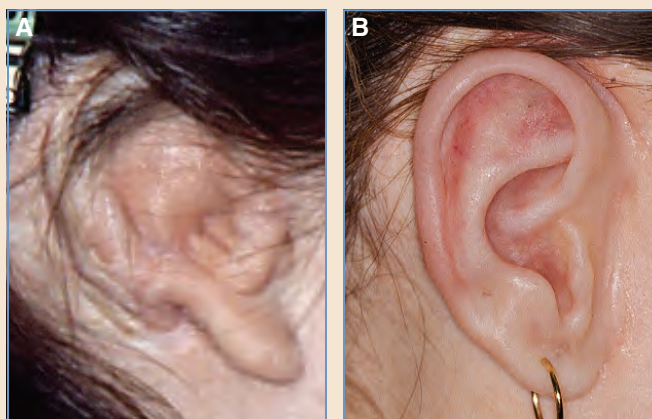


Fig. 31-24 Secondary reconstruction with a prosthesis. **A**, Failed Silastic framework. **B**, Salvage with a prosthetic ear.

5 years for the rest of their lives. Most reconstructive surgeons would much prefer autologous ear reconstruction as the treatment of choice for microtia in children. An adhesive-retained ear prosthesis should not be considered as a trial for an implant-retained prosthesis. It does not accurately demonstrate to the patient the advantage of osteointegration to overcome the significant problems with adhesives (Fig. 31-24).

OUTCOMES

Assessing surgical outcomes is difficult. Perceived results as judged by the patient, family, surgeon, and unbiased observer can be quite different. The psychological outcome of treatment is just as important as the aesthetic result to the patient's overall recovery.

The results of autologous reconstructions have been published.^{32,48-51} Although the overall satisfaction is reported to be high, there are certainly weaknesses in the methods of evaluation. There is no consistent, validated method to evaluate surgical results or to allow comparison of techniques. There is a definite improvement from a psychological point of view.⁵² In 69% of patients, the ear became part of the patient's body image, and 88% would choose autologous reconstruction again; 68% felt the chest scar was "absolutely acceptable."²⁹ Patients younger than 20 years of age at the time of surgery experienced more improvement in self-esteem and psychosocial attitudes with surgical reconstruction.²⁹

Surgical complications such as skin necrosis and hematoma should occur infrequently. These need to be treated early and aggressively to minimize cartilage resorption (Fig. 31-25). Extrusion of a suture is easily corrected by removal. Critics of autologous reconstruction would argue that the results published are only the best results and are not necessarily obtainable by surgeons performing smaller volumes of procedures. However, when the surgery is properly performed by an experienced surgeon, a satisfactory, stable, autologous ear reconstruction should be possible in most cases. The overall experience and training of the surgeon is probably more important than the specific technique used.

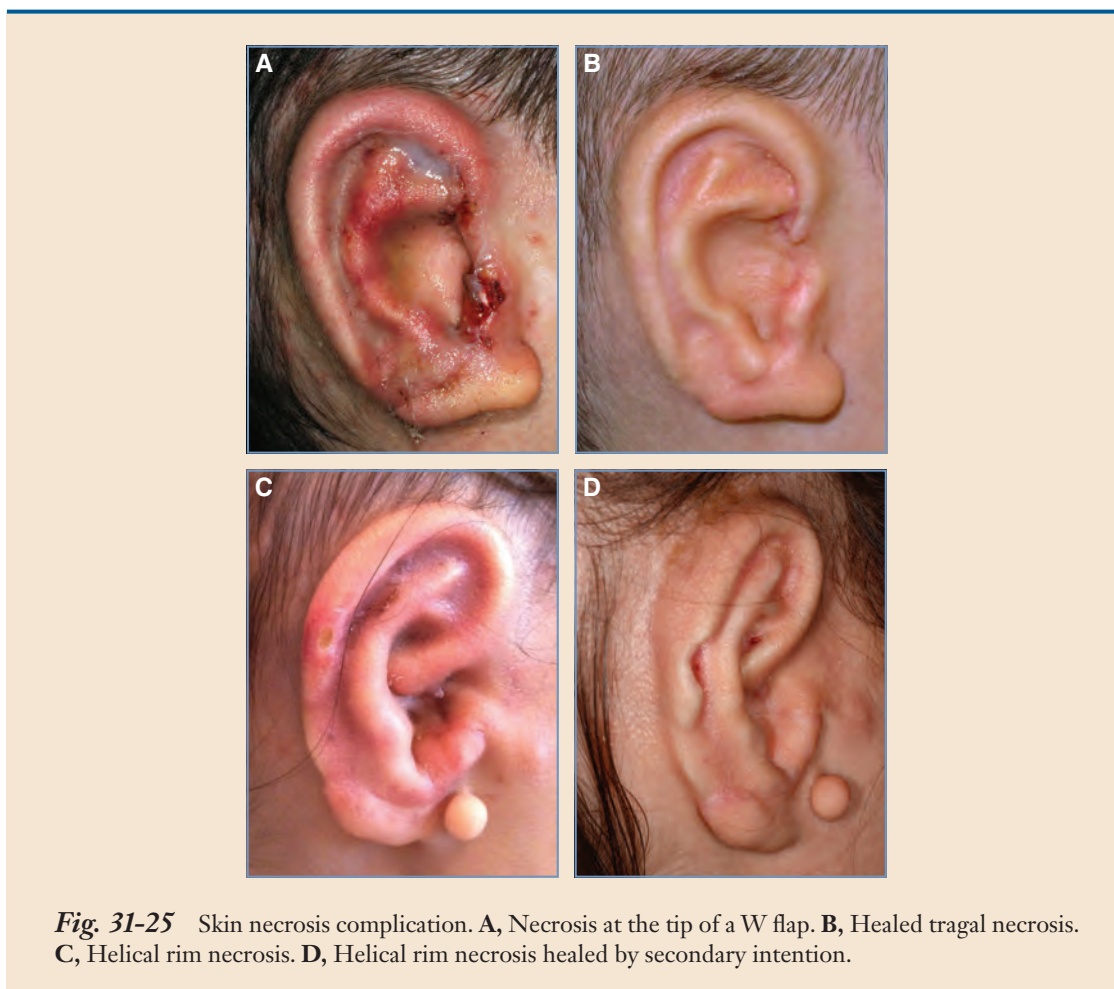


Fig. 31-25 Skin necrosis complication. **A**, Necrosis at the tip of a W flap. **B**, Healed tragal necrosis. **C**, Helical rim necrosis. **D**, Helical rim necrosis healed by secondary intention.

Using porous polyethylene, a high satisfaction rate has also been achieved with an aesthetically acceptable outcome in 85% of children.⁵³⁻⁵⁵ The Glasgow Benefit Inventory (GBI), which measures health-related quality of life, was elevated (21.2) but was not as high as in the autologous group (48.1). Surgical complications decreased when complete coverage of the framework was achieved with a TPF flap. A 2.7% framework fracture rate and a 7.3% exposure rate was reported.⁵³

Osteointegrated prosthetic ear reconstruction has also demonstrated high levels of success in patients of all ages.^{46,47} Psychologically, the prosthesis becomes part of the body image in the majority of patients. Issues related to prosthetic shape, color, means of attachment, and long-term implant stability are minimal. The disadvantages of this approach in children are possible intermittent chronic soft tissue problems around the percutaneous abutments and the ongoing need for a replacement prosthesis.

Tissue Engineering and Composite Tissue Allotransplantation

The future of ear reconstruction rests with advances in tissue engineering.⁵⁶⁻⁶⁴ No material presently available, autologous or alloplastic, completely mimics the characteristics of auricular cartilage. While tissue engineering offers great promise, major hurdles remain. The first problem is

to create a 3D scaffold onto which cartilage cells can anchor and grow. The use of 3D computer-aided design and manufacturing has helped in the creation of very accurate scaffolds. Biological scaffolds mimic cartilage extracellular matrix. Natural materials like hydrogel and hyaluronic acid have been tried but are limited by poor strength and by long-term degradation. Synthetic polymers have the advantage of being customizable: the designer can control the porosity, shape, and permeability of the polymer. However, they do elicit an immunologic response and lack the surface characteristics favoring cellular attachment and growth.

The second issue is populating the scaffold with chondrocytes. One hundred million cells are needed to create an adult ear. Chondrocytes, both autologous and xenogenic, have been used. Auricular and nasal chondrocytes yield more cartilage at a faster rate than auricular cartilage and have superior histologic and biochemical properties. Continued problems remain regarding de-differentiation of cells, limited donor supply, and a short time frame for proliferation. Uncontrolled cell proliferation and potential tumor growth are still concerns. The use of new “smart” scaffolds of nanocomposite polymers with surface modifications to stimulate and control cell attachment, growth and differentiation, and propagation of stem cells may be the way of the future. The problems of skin coverage of the framework and its effect on the ultimate aesthetics will still prove to be a challenge. There have been early reports of use in humans.⁶⁵

The potential for composite tissue allotransplantation in ear reconstruction has been explored from an anatomic perspective.⁶⁶ Long-term challenges of immunosuppression remain to be solved. Its potential use in adults following trauma is much more likely than for correction of microtia in children.

HEARING RESTORATION IN THE MANAGEMENT OF MICROTIA

Ideally, reconstruction of microtia should include both the external ear and restoration of hearing. Many otologists have attempted to restore hearing by creating an external ear canal and reconstructing a tympanic membrane and ossicles to stimulate the normal inner ear.⁶⁷⁻⁶⁹ However, few otologists can consistently restore near-normal hearing.⁷⁰ Complications are high, including frequent restenosis of the canal and recurrent infections. Creating an ear canal risks injury to an aberrant facial nerve. It also causes scar formation in the area of future auricular reconstruction, compromising the final aesthetic result.

The first bone-anchored hearing device (BAHD), introduced in Sweden in 1977,⁷¹ changed the hearing options for children with microtia. These hearing devices are held in position to the mastoid bone by an osteointegrated implant. They allow direct stimulation of the cochlea, which is normal in most microtia patients.⁷¹ Unlike a canaloplasty procedure, this operation does not depend on a functioning middle ear or patent canal and is a much simpler and safer procedure. BAHDs are the benchmark against which newer hearing modalities are measured.⁷²

Bilateral microtia occurs in 10% of patients.²⁵ The significant hearing deficit of these individuals requires early hearing restoration. Our patients are offered the option of trying a BAHD (BAHA Softband) starting in the early newborn period (Fig. 31-26). Surgical placement of a BAHD typically occurs at age 5, when the thickness of the mastoid bone is sufficient for a titanium implant. The most common surgical practice is to place a unilateral BAHD, even in bilateral microtia, because a single hearing aid will stimulate both cochleas simultaneously. Placement of bilateral BAHDs is being investigated⁷³; recent evidence suggests that this results in better hearing sensitivity, speech perception, sound localization, and lateralization, as well as improved patient perception of overall sound quality and quality of life.⁷⁴⁻⁷⁷

The application of a BAHD in unilateral microtia patients is less clear, since there is a commonly held belief that these patients do not require hearing restoration. However, there is con-



Fig. 31-26 OAVS patient wearing a BAHA Softband hearing device.

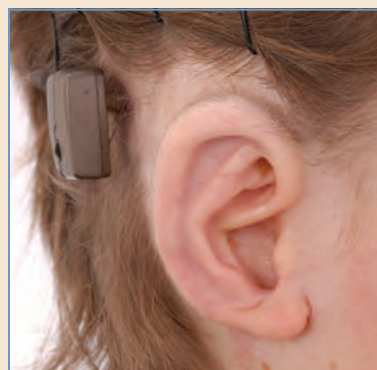


Fig. 31-27 Autologous reconstruction with BAHD.

siderable evidence to suggest that untreated unilateral hearing loss causes deleterious effects to the education and behavioral development of these children. This warrants reevaluation of traditional practice and study of hearing restoration with a BAHD.⁷⁸⁻⁸⁴

Advances in percutaneous, transcutaneous, and completely implantable hearing devices will give patients with microtia new hearing options.⁸⁵⁻⁸⁸ The long-term success and effects of such devices on autologous reconstruction still requires further evaluation. The ability to upgrade and change the hearing hardware in the future needs to be considered when discussing current treatment options with the patient and their families.

Autologous Ear Reconstruction and Bone-Anchored Hearing Devices

A team approach is needed to satisfy the needs of both the otologist and the reconstructive surgeon. Appropriate placement of the bone-anchored hearing device is crucial both for hearing and to avoid scarring at the future site of autologous reconstruction.⁸⁹ The distance the bone-anchored hearing device should be sited from the pseudoauditory meatus is not well established. Our placement has averaged 5.6 cm from the pseudomeatus. There was no significant difference in positioning or complications whether or not the implant was placed before or after autologous ear reconstruction (Fig. 31-27).

Another challenge is the timing of each operation. The pediatric otologist would prefer that the bone-anchored hearing device be implanted before school age (around age 5) to prevent any

deleterious developmental effects of hearing impairment. The plastic surgeon would prefer that the autologous reconstruction be done before implant placement (age 8 to 10) to decrease the risk of skin complications or future ear-positioning issues.

LEARNING TO PERFORM MICROTIA RECONSTRUCTION

One of the reasons for the inconsistency in surgical results has been related to challenges in training surgeons to do the procedures.⁹⁰ Traditional surgical teaching has usually included some form of observership but has lacked “hands on” experience. Costal cartilage from the sixth, seventh, and eighth ribs presents unique challenges of shape, form, and consistency that are not captured in traditional training models.^{91,92}

Better hands-on training simulations, including rib and multipiece ear framework models, have become available to test the surgeon’s aptitude, commitment, and skill level⁹³⁻⁹⁵ (Fig. 31-28). The authors of this chapter have developed an app available at iTunesU (<http://www.apple.com/education/ipad/itunes-u/>) to teach the intricacies of ear carving using a step-by-step approach

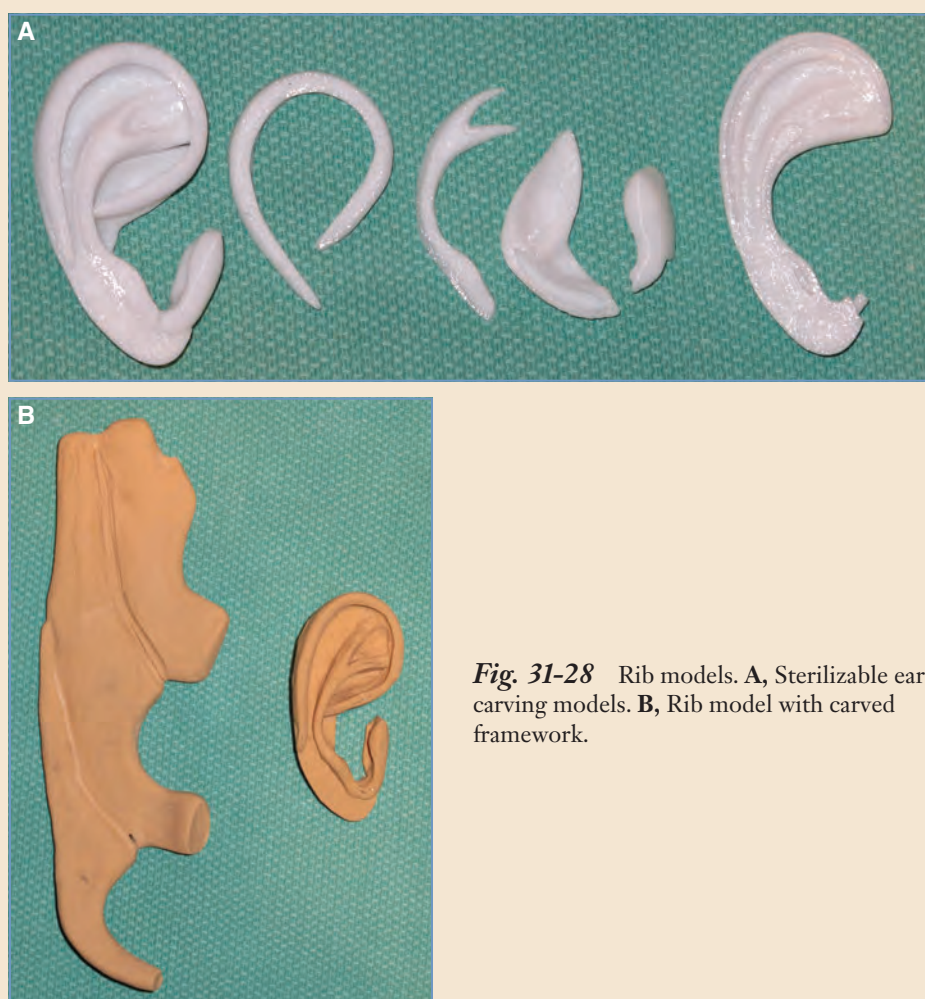


Fig. 31-28 Rib models. **A**, Sterilizable ear carving models. **B**, Rib model with carved framework.



Fig. 31-29 iPad application for teaching microtia reconstruction.

(Fig. 31-29). This application aims to shorten the learning curve for surgeons. It will also dissuade those unable to create an accurate three-dimensional ear framework on a model from actually operating on a patient. Also available are several surgical videos by leaders in ear reconstruction demonstrating their techniques to create an ear framework from rib cartilage.^{16,18}

Surgical simulation and the use of models are playing an increasing role in surgical education.⁹⁶ Surgical simulation studies particularly in general surgery and urology have led to the development of four principles we think are applicable to microtia training:

1. Mandatory participation
2. Proficiency-based learning (not simply time or numbers based)
3. A distributed training schedule (not a weekend course)
4. Overtraining

These concepts have been applied to athletes, musicians, and pilots and would likely benefit reconstructive ear surgeons as well. Simply watching an experienced surgeon perform ear reconstruction is not a substitute for practiced skills. It is no longer acceptable to just “give it a try” if the surgeon has not demonstrated some proficiency using an ear-carving training model or its equivalent.

This sensible approach to surgical education will result in fewer but better trained reconstructive surgeons. They will be performing autologous ear reconstruction with better and more consistent outcomes.

CONCLUSION

Ear reconstruction continues to be a challenging, dynamic field of reconstructive surgery. Results will improve only with improved educational efforts and specialized surgeons offering this form of reconstruction. Developments in tissue engineering and advanced digital technologies offer the greatest hope for future improvements in the care of patients with microtia.

KEY POINTS

- There are several classification systems used to describe microtia based on the degree of ear deformity. The more descriptive the classification system, the more relevant and useful it is for surgical planning.
- Microtia is often associated with an identifiable syndrome. Clinicians should be aware of this so that a thorough workup is performed to rule out additional anomalies.
- There are three methods currently used for ear reconstruction: autologous reconstruction with costal cartilage, reconstruction with a porous polyethylene framework (such as Medpor), and an osteointegrated implant-retained prosthesis.
- Autologous reconstruction using costal cartilage has evolved over time. The most recent techniques require more cartilage to produce a more elaborate 3D framework.
- Care must be taken to confirm the correct position of the newly constructed ear based on anatomic landmarks and facial features. A poorly positioned ear will yield a suboptimal result regardless of the appearance of the ear itself.
- The costal cartilage framework can be carved based on templates traced from the normal contralateral ear, in the case of a unilateral microtia, or it can be carved based on prefabricated templates designed by Nagata.^{7,8}
- In general, the sixth, seventh, and/or eighth ribs are harvested for construction of the 3D framework. Resultant chest wall deformity can be mitigated by reintroducing the diced leftover cartilage into a pseudorib of resorbable mesh and placed in the perichondrial sleeve.
- Regardless of the technique used, delicate handling of soft tissue during reconstruction is imperative. The framework, be it cartilaginous or synthetic, must be covered with healthy, well-perfused tissue in order to prevent exposure of the framework. For porous polyethylene (PPE or Medpor) reconstruction, the framework must be covered in its entirety with temporoparietal fascia.
- Most autologous methods of reconstruction require more than one stage. Elevation of the framework is generally reserved for a second or later stage to avoid undue tension on the skin pocket at the primary procedure, which would risk skin necrosis.
- Autologous ear reconstruction is more technically difficult in syndromic patients (especially those with mandibular hypoplasia), low-hair-bearing patients, and in secondary cases.

Continued

KEY POINTS (continued)

- Each type of ear reconstruction has its own individual merits. The mode of reconstruction chosen should be based on patient factors as well as surgeon experience.
- If restoration of hearing is being considered, there needs to be careful communication and planning by all surgeons involved to ensure that the procedures do not interfere with one another in terms of aesthetic result and function.
- All of the reconstructive techniques described in this chapter can produce a pleasing aesthetic result when performed by an experienced surgeon. However, gaining this experience is difficult; it requires mentorship and practice. Surgical models have been developed to allow the inexperienced surgeon to learn the skill of carving outside of the operating room. We highly recommend that surgeons interested in learning ear reconstruction take the necessary time and steps to become proficient before embarking on a reconstructive ear case.

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32

Nonmicrotia

Bruce S. Bauer



Bauer Otoplasty Technique.



As varied as individuals are, ears vary in their size, prominence, and shape. These variations are recognized as “normal” as long as the size of the ear is in balance with the other facial features and in a correct relationship to them.¹⁻³ In this increasingly visual world, when the media wish to depict an oddball character, a less intelligent individual, or a “funny-looking kid,” they often select subjects with large, prominent, or oddly shaped ears. Regardless of what we may later learn about these characters, we initially assign a level of immaturity or low intelligence to them. Even minor disturbances in balance or minor abnormalities in shape are immediately perceived as looking “wrong,” and this perception can become the source of significant peer ridicule. Macgregor⁴ noted, “For the child with microtic, protruding, or lop ears, for instance, such an abnormality may be the source of intense shame and anguish.” These concerns often persist well into adulthood if the deformities are not corrected, despite the fact that as an adult the individual may have developed good coping mechanisms. Not surprisingly, altering these abnormal features to a size or shape that “looks right” can have a remarkably beneficial effect on a child’s self-esteem and body image.^{2,4}

Given the well-established fact that the ears reach near-adult size and proportions relatively early in the overall scheme of facial growth and development,⁵ the previously described concerns underscore the need for timely evaluation and treatment. Although I do not advocate early correction of ear deformities to appease overanxious parents, we must recognize that it is not uncommon to see adult patients who present for correction of congenital ear deformities who wish that their parents had allowed these deformities to be corrected when they were young children. It is therefore essential that, when considering the timing of ear reconstruction, we balance what we know about ear and facial growth, available donor tissue for reconstruction, and the capabilities of children to both tolerate a corrective procedure and follow the appropriate postoperative routines.

In this chapter I describe my approach to both the analysis and treatment of the more common nonmicrotia ear deformities, and I lay the groundwork for applying these techniques to rare and unusual deformities that come along. Discussing the techniques used to treat congenital deformities has a direct bearing on treating acquired deformities, something that will become quite apparent in the following chapter. Although I will review the common numbers and measurements given for normal ears, I think that one should concentrate on recognizing the elements of protrusion, excessive size, distortion of parts, and position abnormalities that require correction to establish an appearance for a reconstructed ear that appears “right.”

EAR ANATOMY

The external ear consists of a three-tiered cartilaginous framework with adherent skin on its anterior surface and more loosely applied skin on its posterior surface. The three tiers of delicate cartilage form the helical-lobular complex, the antihelical-antitragal complex, and the conchal complex. The normal complete ear includes the scapha, concha, helix, antihelix, tragus, and lobule. The antihelix, antitragus, lobule, and Darwin's tubercle may be absent or poorly developed, yet distortion or absence does not always result in an abnormal-appearing ear^{1,2} (Fig. 32-1).

Although there are wide variations in the proportions, projection, and position of the ears among individuals, and even in a single individual, it is important to have some guidelines for establishing the degree of abnormality present. Tolleth¹ summarized the more pertinent numbers, and these are reviewed here (see Fig. 32-1).

- *Axis:* The line passing through the longest dimension of the ear is the axis. Although it is often considered to lie parallel to the nasal bridge, an angle of 20 degrees between these structures is more correct in most people. It is most important to recognize that the axis of the ear is rarely vertical.
- *Position:* When the head is in an anatomically vertical position, the top of the ear should be on a line with the top of the brow, and the bottom of the ear should be on a line at the base of the columella or slightly lower. The ear is positioned approximately one ear length (6.5 to 7.5 cm) posterior to the lateral rim of the orbit.
- *Size:* Wide variations also exist in the standard methods of measuring ear size and the consequent measurements. Measuring the ear height along the long axis and the width at right angles to that, the ear varies in height from 5.5 to 7.5 cm and in width from 3.0 to 4.5 cm (50% to 65% of the height).
- *Protrusion:* Although measurements and standards can be listed in both degrees and centimeters of protrusion, many surgeons find an actual centimeter measure most practical in the operating room. A measurement of 1.5 to 2.0 cm from the scalp to the anterior surface of the superior pole of the helix most commonly provides a visually correct degree of protrusion.^{1,2}

Optimal aesthetic reconstruction of all congenital auricular deformities is based on a clear understanding of both the cartilage and soft tissue anatomy of a normal ear and a deformed ear and on an appreciation of the skeletal asymmetries that often coexist with these deformities. Although it may be obvious that there is an association between microtia and hemifacial microsomia, the effects of other skeletal anomalies (that is, positional, nonsynostotic plagiocephaly) on the position and prominence of an ear may be considerably more subtle. A reconstructed ear must not only be the correct size and shape but also bear the correct relationship to other facial features. The relationship between ear position and conformation of the underlying skeletal supporting structures is an important theme in the discussion of both prominent ears and the reconstruc-

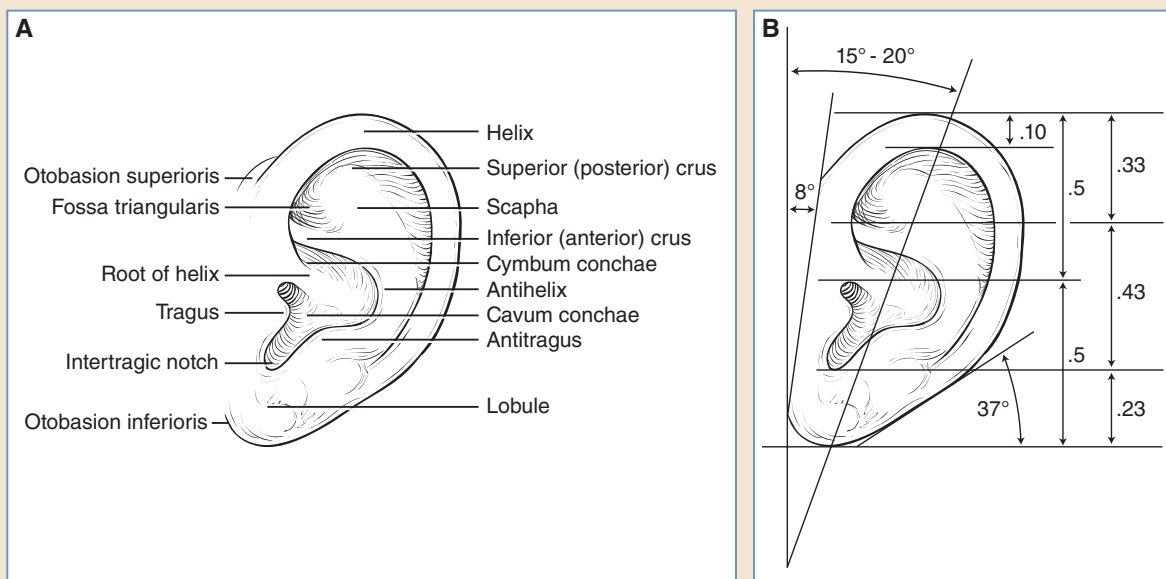


Fig. 32-1 A, The normal ear and its parts. B, The ear's critical proportions.

tion of microtia.^{6,7} One must also be aware that variations in cranial vault shape may influence ear position and prominence, and this may throw off standard measurements. I am convinced that one should correct ear protrusion to what looks right rather than to a specific centimeter measurement.

TIMING OF SURGERY

There are three main considerations for determining the optimal timing for reconstructive procedures of the ear:

1. Psychological—timing the reconstruction to avoid undue psychological trauma from the deformity
2. Anatomic—taking into consideration the size of the contralateral ear in unilateral deformities and the strength of the available auricular donor cartilage in both unilateral and bilateral reconstructions
3. Growth—considering the growth of both of the ears in general and the growth of the ear after reconstructive procedures

Adamson et al⁵ reviewed the growth patterns of the external ear and concluded that (1) the ear reaches 85% of its adult size by the time a child is 3 years of age, (2) growth continues until adulthood, but little change in width or distance from the ear to the scalp occurs after 10 years of age, and (3) for all practical purposes, the normal ear is almost fully developed by 6 years of age. Farkas^{8,9} differed somewhat in his measurements, stating that the ear reaches 85% of full size by 8 years of age, 90% by 9 years, and 95% by 14 years. Work in the 1980s clearly showed that there is continued ear growth in adulthood, although conchal width does not change appreciably. This is something we intuitively recognize in viewing many elderly adults. This may play a factor in

deciding on whether to reduce the ear size when applying the varied otoplasty techniques that we will discuss with the adult patient.

I have had enough experience with unusual cases, in which I felt correction before 5 years of age was appropriate, to comfortably state that early surgery does not affect the long-term growth of the ear. However, there are practical considerations that argue against routine early intervention. Few if any children express concern about the abnormal appearance of their ears before reaching 5 or 6 years of age, even when the deformity is significant enough to elicit comments from their peers.^{10,11} The proportionally greater growth of the ear in these early years in relation to the rest of the facial features may in fact accentuate the prominence of the ear. Correction before this age, although quite possibly easing parental concerns about the child's appearance, may complicate the postoperative course if the child, even if not intentionally, pulls off the bandages and potentially disrupts the repair. Nevertheless, I have seen children who were clearly aware of their appearance by 4 years of age and were bothered enough that they both eagerly anticipated surgery and cooperated totally during the healing period.

Regarding the more complex nonmicrotia deformities such as group II and III constricted ears, Stahl's ear, and some cryptotias, surgery may need to be delayed until the conchal cartilage used for "expanding" the auricular framework is sturdy enough to support the changed skin envelope. This is certainly the case by 5 or 6 years of age. There are occasional group II constricted ears where unfolding the vestige along with posterosuperior reposition may help both with hygiene of the canal and masking of the more significant deformity until the age where the greater part of the reconstruction or otoplasty can be accomplished; however, this is more of an issue in microtia cases associated with significant displacement of the vestige because of underlying skeletal asymmetry.

PERIOPERATIVE CONSIDERATIONS

There are several perioperative considerations, such as prophylactic antibiotics, surgical preparation, and draping techniques, that should be discussed briefly here before proceeding with the specifics of the different ear deformities. There is considerable variation as to how each of these considerations is addressed from one surgeon to another, so these are included among a selection of several approaches.

Prophylactic antibiotics are recommended for most aesthetic and reconstructive procedures of the ear. For most surgeons, the risk of chondritis, the most feared complication of otoplasty, is a sufficient indication that at least an intraoperative dose of a broad-spectrum antibiotic should be given, and most surgeons also prescribe a similar oral agent for the first few postoperative days. Few surgeons allow sufficient time to thoroughly prepare the auditory meatus and canal with Betadine or a similar iodinated preparation (a routine strongly advocated by Brent¹²), even though this might be sufficient enough to eliminate the risk of chondritis. In my view, active ear drainage is a contraindication to all surgery on patients with nonmicrotia deformities. However, if a child has had myringotomy and tube placement and has some typical drainage, this is not necessarily also a contraindication.

Surgical draping is also subject to considerable variation based on each surgeon's preference. I prefer a standard head drape with staples to hold the drapes in place, which allows a full view of the face and both ears. This exposure is particularly important in patients with significant asymmetries. In addition to the head drape, two halves of a medium piece of Tegaderm cut lengthwise provide an excellent barrier to keep sideburn hair and postauricular hair out of the surgical field. Another alternative is using a single larger piece of Tegaderm over the whole area surrounding the ear, with a hole cut out for the ear.

RECONSTRUCTIVE TECHNIQUES FOR CONGENITAL NONMICROTIA AURICULAR DEFORMITIES

Prominent Ears

Anatomy and Goals of Otoplasty

The prominent ear is traditionally characterized by two major elements but is also influenced by additional factors. First, there is usually some degree of effacement of the antihelical fold, with an increased scapha-conchal angle causing increased projection of the superior pole of the ear. Second, the majority of cases show some degree of hypertrophy of the concha, which results in displacement of the entire ear structure from the side of the head. Ear position is also influenced by the shape of the skull. If there is preexisting plagiocephaly, asymmetry between the ears can be accentuated by the steeper underlying skeletal plane on one side compared with the other. In addition, upper pole prominence is accentuated in patients with a heart-shaped face in which there is a broader intertemporal distance than in an individual with a narrow face and mandible. The technique for correction of ear prominence needs to respect the effect of the underlying skeletal plane on both the upper and lower poles of the ear to ensure complete correction of the deformity. Without a degree of overcorrection of the upper pole, the end result may appear undercorrected. Techniques need to be further modified to correct macrotia and overgrowth of the lobule.

The most important factor in the correction of prominent ears, as well as the more complex deformities mentioned in this chapter, is correct analysis of the deformity and recognition that quite different factors may be at play on each side (Fig. 32-2). Most otoplasties will include some



Fig. 32-2 Variations in anatomy and causes of ear prominence. **A**, Significant prominence resulting from near-total effacement of the antihelical folds (with a suggestion of macrotia). **B**, Prominence resulting from conchal hypertrophy. **C**, Prominence in association with hypoplasia of the underlying craniofacial skeleton. **D**, The critical angle of cartilage between the conchal floor and antihelix influences the prominence of the lobule.

reduction of the concha, correction of the effaced antihelical fold, and additional elements, from reduction of ear size to the contour of the helical rim, antihelix, and lobule. Again, although normal ear proportions have been well documented, the recognition of abnormal size and projection is so innate that one's sense of normal is a much better guide in otoplasty than actual measurements.

Antihelical Fold

Effacement or deficiency of the antihelical fold ranks first in most discussions of the prominent ear.¹³⁻¹⁷ This deformity presents as a spectrum ranging from a totally indistinguishable antihelix with a confluent concavity from the antihelix to the scapha and the helical rim projected outward and forward, to a loss of definition of only the superior antihelix with prominence of the upper pole of the ear.

Conchal Size and Shape

Although less attention has been given to the importance of the concha in overall shape and projection of the ear,^{7,18,19} it makes sense, when thinking about the three-tiered configuration of the auricular cartilage framework, that the delicate antihelix and helical complex are mounted on the sturdier concha so that changes in conchal size and shape profoundly influence the overlying tiers. In my experience, it is rare to see ear prominence that does not have a conchal element.

There appear to be three ways in which the concha affects the prominence of the ear (Fig. 32-3):

1. Overall enlargement of the concha projects the ear away from the mastoid surface.
2. An extension of the helical crus across the concha creates a firm cartilage bar that pushes the ear outward.
3. The effect of the cartilage angulation at the junction between the cavum concha and the sweep of cartilage up to the antitragal prominence affects the position and prominence of the lobule and lower third of the ear.

The first element is well recognized. Little attention has been given to the second element, but once it is seen, it is easy to understand. The third element, once recognized, opens the door for understanding the approach to the isolated lower pole and lobule prominence.

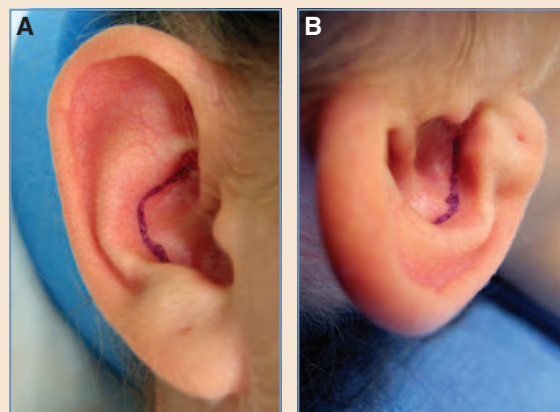


Fig. 32-3 Combined otoplasty technique. **A** and **B**, Two views of the conchal incision.

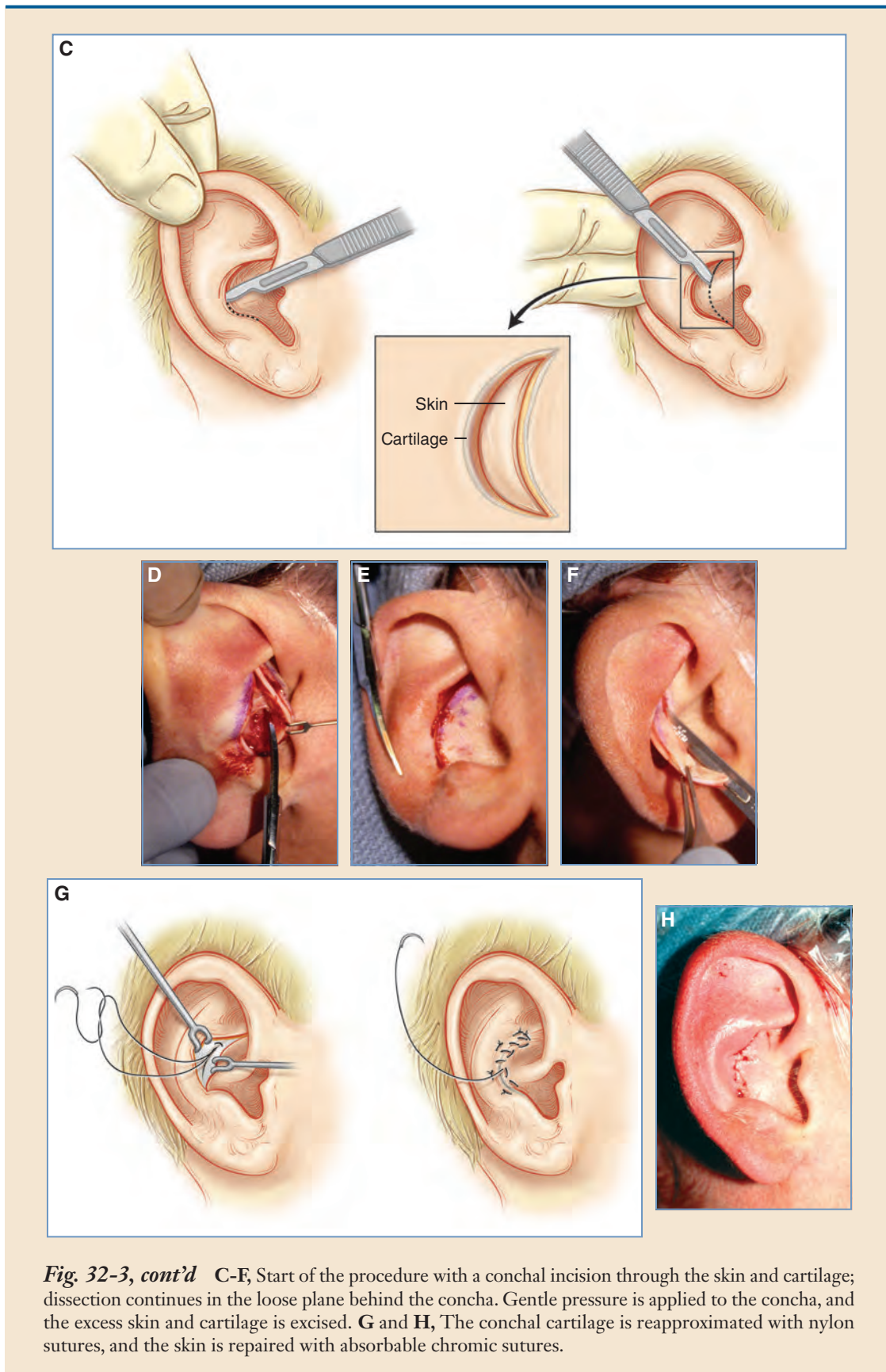


Fig. 32-3, cont'd C-F, Start of the procedure with a conchal incision through the skin and cartilage; dissection continues in the loose plane behind the concha. Gentle pressure is applied to the concha, and the excess skin and cartilage is excised. G and H, The conchal cartilage is reapproximated with nylon sutures, and the skin is repaired with absorbable chromic sutures.

Continued

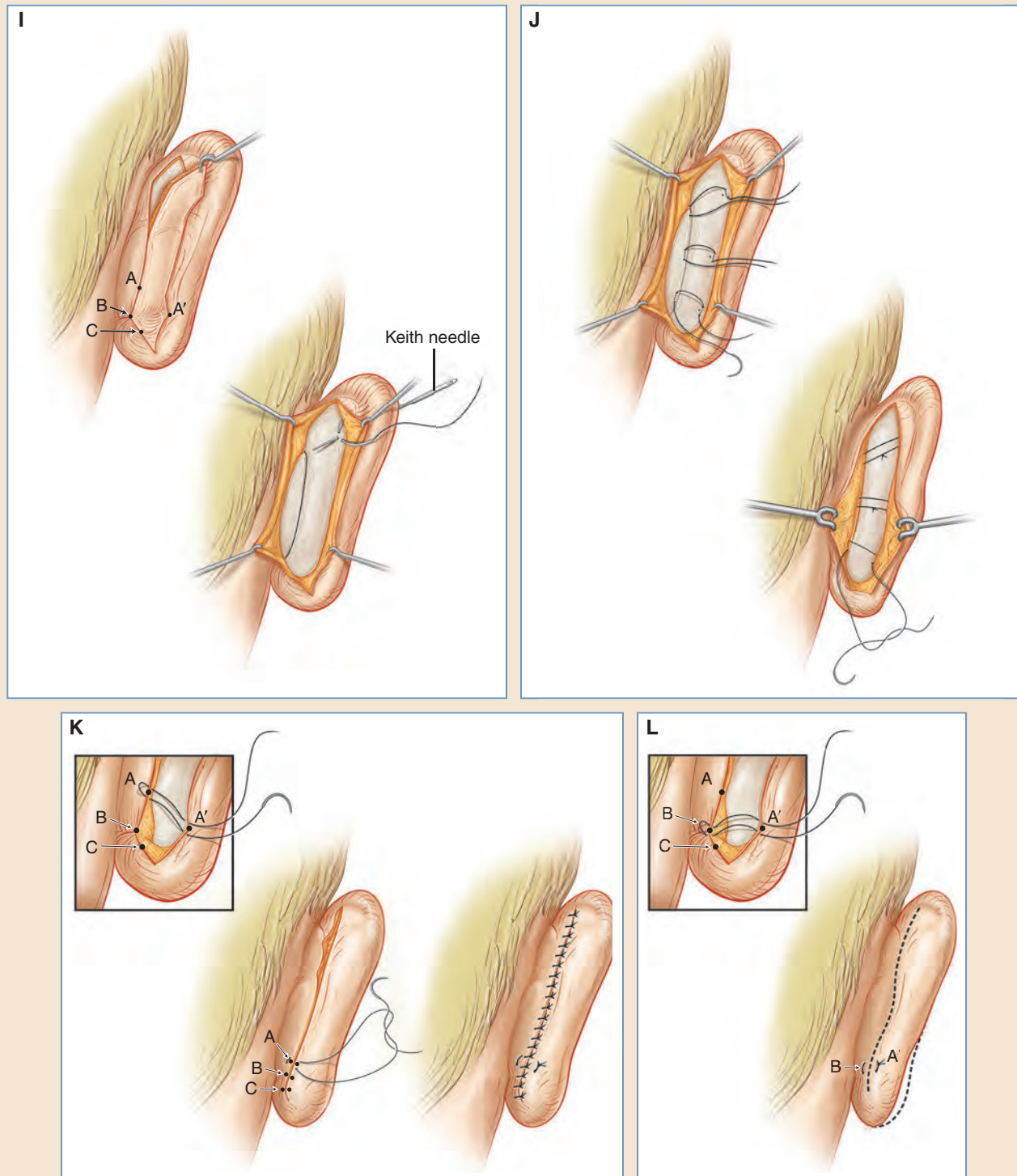


Fig. 32-3, cont'd I and J, The squid-shaped skin excision is followed by exposure of the perichondrium on the postauricular surface. The straight Keith needle marks the position for placing scapha to mastoid, helical sulcus to mastoid, and additional sutures, and these sutures are tied to adjust prominence. K and L, The final position of the lobule is set with placement of the first closing suture between point A' and points A, B, or C. The different positions of the sutures will vary the shape of the lobule as illustrated.

The third element of conchal shape, although it is not the only cause of lobule prominence, appears to play a key role. As the cartilage angle between the concha cavum and the antitragus becomes more acute (that is, as the antitragus tips closer toward the concha), this supporting structure projects the lower third of the ear and lobule outward. I think that this feature has a greater influence on the lobule position than the commonly described helical tail.

Anatomy of the Underlying Skeleton

Little attention has been given to the effect of the underlying skeleton on ear prominence, but it should be recognized and taken into account when planning the correction of prominent ears. The external ear is mounted on the bony base of the underlying temporal bone. Anomalies in skeletal shape and skeletal asymmetries can affect one or both ears. Perhaps the most recognizable example of this relationship is the change in ear position and projection in association with positional, nonsynostotic plagiocephaly. With the parallelogram deformation of the cranial vault, the ear on the side of occipital flattening is projected forward and is often more prominent.

A less subtle example of the influence of the skeleton on ear position is the effect of deficient temporal bone and medial positioning of the temporomandibular joint in hemifacial microsomia. In severely affected cases, without microtia, the normal ear may seem to be “sheared off the head,” with the upper half of the ear projecting outward and the lower half canted medially toward the hypoplastic face (see Fig. 32-2, C). Another example that shows some similarities is those patients whose general craniofacial shape includes a relatively broad head with a narrow face and mandible. Seen from the front, the face and head have a heart-shaped configuration. This slope from head to face may project the upper ear outward, creating ear prominence but showing otherwise normal and proportional ear features.

Although otoplasty techniques vary from one case to another, the overall goals of otoplasty remain the same¹³:

- Correction of protrusion (particularly the upper pole)
- Visibility of the helix and antihelix
- Smooth antihelical line
- Undisturbed postauricular sulcus
- Avoidance of a “plastered-down” look
- Avoidance of a sharp antihelical fold

Correction of a Prominent Ear

A review of all or even most otoplasty techniques is beyond the scope of this chapter. I will instead attempt to review the techniques that have been emphasized most often in the literature for correcting the typical prominence caused by insufficient formation of the antihelical fold and those cases resulting from conchal hypertrophy. Techniques must be varied when both elements of prominence require treatment and when overall reduction in ear size is also required. After a brief overview, I will discuss the approach that has evolved over my more than three decades of ear surgery. This approach has the greatest flexibility for correction of prominence in the full spectrum of nonmicrotia ear deformities and how the correct sequence of these procedures will lead those less familiar with this surgery to a significantly better understanding of the problem and how even subtle deformities can be corrected with high certainty of success.

Creating the Antihelical Fold

A review of both the plastic surgery and otolaryngology literature reveals a multitude of techniques for correction of the antihelical fold.^{13,15-22} In general, these techniques can be grouped into those that score or abrade the cartilage to control the direction and extent of folding and

those that rely on suture fixation of the cartilage.¹⁸⁻²⁰ In addition, there are a number of procedures that combine these techniques.^{6,7,20,21}

The cartilage-scoring techniques vary, based on whether the cartilage is simply scored²⁰ or actually cut through,²² and when it is scored, whether the scoring is on the anterior or the posterior surface of the cartilage. Although any of these techniques can be successfully accomplished by skilled hands, the risk of a sharp antihelical fold is considerably higher when a full-thickness cut is used in the body of the antihelical cartilage. Keeping in mind the potential dissatisfaction most patients will express when the cartilage shows surface irregularities and sharp, unnatural folds, I think that scoring techniques should be relegated to the history books—particularly given the fact that other techniques are available that easily avoid this problem, and all degrees of effacement can be corrected without the need of any disruption of the smooth surface of the antihelix.

Conchal Reduction

Some degree of conchal hypertrophy is present in virtually all cases of ear prominence, although certainly it is less significant in some cases. In my experience the largest number of patients who are dissatisfied with the results of a previous otoplasty focus on the overaccentuation of the antihelical fold to correct prominence when conchal hypertrophy has been missed or ignored as an important element of their initial prominence.^{7,21}

The techniques for correcting conchal hypertrophy, once properly identified, include lowering the concha by creating a new antihelical fold, conchal setback with conchal mastoid sutures, and conchal reduction.^{7,15,21} The reduction procedures can be divided into those that excise cartilage alone (usually through a posterior approach) and those that resect both cartilage and skin through an anterior approach. Although minor degrees of conchal prominence are handled through the creation of a new antihelical fold or by using a conchal mastoid suture, I think that most patients with significant degrees of conchal hypertrophy require cartilage excision.^{7,21}

The choice of a posterior or anterior approach to the cartilage excision is, for the most part, one of personal preference; either approach is effective, and a surgeon gains facility with the approach used most frequently. In my opinion, there is considerably more control with an anterior approach than a posterior one, and in cases where the resection will be 0.5 cm or more in width, the extra skin left from the use of a posterior approach may not shrink down sufficiently to avoid leaving a visible fold in the conchal floor. This fold may be more visible than a fine anterior conchal scar, which is rarely visible by 1 year after otoplasty. In cases in which a conchal incision is used to help break the spring of the antihelical body to allow smooth reshaping and no conchal resection is planned, the cartilage incision can be made from a posterior approach alone, although these cases are relatively few. With meticulous skin closure, the scar is rarely visible and in my many years of ear surgery both for correction of nonmicrotia deformities as well as harvest of composite conchal grafts, I have seen only a single patient with hypertrophic scarring in the concha (a CHARGE* syndrome patient).

Current Combined Otoplasty Approach

The evolution of my current technique was from early use of the technique described by Stark and Saunders,²⁰ which combined posterior thinning (dermabrasion) of the proposed antihelical fold and suture fixation, with very rare conchal reduction. In the cases where some degree of

*CHARGE stands for *coloboma*, *heart defect*, *atresia choanae* (also known as *choanal atresia*) *retarded growth and development*, *genital abnormality*, and *ear abnormality*.

conchal reduction was carried out, it was immediately evident that there was much better control of the folding of the antihelical fold and little or no need to thin the antihelical cartilage to get it to bend while the sutures were tied to guide the correction of the antihelical effacement. Even with careful posterior thinning of the antihelical cartilage, some irregularities were seen on the anterior surface. Combining these observations with those from a group of secondary otoplasty patients in whom the initial otoplasty surgeon clearly failed to recognize the significant conchal hypertrophy underlying their deformities and in compensation sharpened the antihelical fold to the point of losing all visibility of the helical rim, a technique centered on “remodeling” the concha seemed to be the best match for the anatomy.

Otoplasty with chondrocutaneous conchal resection as the cornerstone of correction, in combination with posterior suture shaping and fixation of the antihelical fold, avoids any thinning or potential irregularities in the antihelix that can occur with many of the cartilage-scoring techniques, and minimizes the risk of recurrence that may be seen with suture techniques alone. The integrity of the antihelical cartilage is maintained, because the perichondrium overlying both surfaces of the antihelix is left undisturbed, yet the cartilage spring is diminished as a result of the “break” at the conchal level.

With the exception of rare cases in which there is no visible conchal component to the deformity, the procedure begins with a conchal incision and excision of a crescent of cartilage with a smaller skin component (see Fig. 32-3, *A* through *H*). For lesser degrees of cartilage resection, no skin should be excised. After repair of the concha, attention is turned to the postauricular surface, where skin excision and exposure of the posterior perichondrium over the scapha, helical sulcus, and helical tail is followed by suture placement. Sutures are tied to correct prominence, and closure of the inferior incision sets the shape and correction of the lobule (see Fig. 32-3, *I* through *L*). With these steps completed, adjunctive procedures may be performed, such as correction of helical rim deformities, correction of excessive antitragus prominence, and additional reduction of lobule size. Although one may feel that the foregoing discussion of the shift in thinking about and approaching the prominent ear in general and concha specifically is time consuming, it is critical for surgeons to understand and correct the whole range of nonmicrotia deformities, both classic and rare.

Surgical Technique

Details of the procedure follow, but the preceding general sequence should be kept in mind. Although I have made every effort to describe the nuances of each step, the indications and benefits of each maneuver become apparent as one gains experience with the procedure and a better appreciation of the flexibility inherent in this approach. With the rare exceptions noted, the techniques used are similar for both children and adults.

With the patient under general anesthesia using a reverse-airway endotracheal tube (or under intravenous sedation), the face and both ears are sterilized with a green soap solution and subsequently draped with a standard head drape. Stray hairs are secured with Tegaderm or some other clear, sterile drape. A solution of 0.5% lidocaine with 1:200,000 epinephrine is infiltrated into the anterior and posterior surfaces of both ears. Bilateral great auricular nerve blocks with 0.25% bupivacaine are also injected for postoperative analgesia. A broad-spectrum antibiotic is administered through an intravenous line.

The procedure typically begins on the more prominent side, with the incision on the anterior surface of the concha. The incision is placed at the junction of the posterior conchal wall and the floor of the concha (see Fig. 32-3, *A* through *C*), beginning in the cymba concha and continuing along the cavum concha to a point below the antitragus but not as far as the external auditory

meatus. This incision is deeper in the concha than the incision described by Elliott.¹⁵ Placing the incision too high in the concha decreases control of the antihelical fold, allowing the cut edge adjacent to the antihelix to spring forward when the antihelix is shaped. The incision is carried through both the anterior skin and conchal cartilage, stopping shy of the posterior conchal skin. Sharp dissection in the loose areolar plane behind the concha (not in the subperichondrial plane) frees soft tissue and skin from the posterior surface of the conchal bowl (see Fig. 32-3, *D*). A gentle setback of the antihelix with finger pressure accurately estimates the degree of conchal hypertrophy and the amount of cartilage and skin resection needed to create an aesthetically pleasing position for the ear (see Fig. 32-3, *E*).

Usually a crescent-shaped chondrocutaneous segment is excised (see Fig. 32-3, *C* and *F*). To ensure a tension-free closure, more cartilage is typically resected than skin, particularly along the inferior concha deep to the antitragus (where excessive resection can pull the antitragus upward and increase the prominence of the lower pole of the ear and lobe). The edges of the conchal cartilage are approximated with three or four interrupted 5-0 clear nylon sutures (see Fig. 32-3, *G*). How the inferiormost suture is placed at the junction of the cavum concha and antitragus adjusts the angle between these landmarks and strongly influences the cartilage support to the lobule. A slight overlap of cartilage (antitragal segment over conchal segment) tips the lobule posteriorly. Once the cartilage is repaired, the anterior skin is closed with a combination of 5-0 chromic gut with horizontal mattress sutures, followed by a running 6-0 mild (fast-absorbing) chromic gut suture (see Fig. 32-3, *G* and *H*). With these maneuvers completed, the helical rim and ear might seem more prominent at first, but gentle pressure again applied to the antihelix demonstrates a relaxing of the tension needed to bend the antihelix back for its desired correction.

Correction of an effaced antihelix and prominence of the lobule begins with a retroauricular squid-shaped skin excision down to the perichondrium. The squid-shaped excision (which, in essence, is the typical dumbbell-shaped ellipse with the inferior end widened to a diamond shape) is designed to allow access to the posterior surface of the scapha and helical sulcus for suture placement and to assist in correction of the lower pole prominence. The diamond-shaped inferior extension of the squid is positioned with its maximal width at the point of maximal lobule prominence (see Fig. 32-3, *I*). Exposure of the posterior surface of the conchal bowl and scapha is achieved without dissection of any perichondrium off the cartilage.

Preserving the perichondrium is essential to lessen the possibility of subsequent sutures pulling through the cartilage. As the dissection proceeds to expose the posterior surface of the cartilage, care is taken when dissecting the area of the helical sulcus to identify the helical tail. The latter is seen as a duplication of cartilage extending inferiorly from the middle to lower third of the helical rim. This dissection must not free the cartilage of the tail from the skin overlying its anterior surface, because this would diminish the effectiveness of the correction of lobular prominence. The final dissection proceeds onto the mastoid and temporal surfaces, freeing the soft tissues sufficiently to ensure that later sutures can be securely placed into the fascia rather than more mobile overlying tissue. Bipolar cautery is used throughout this process to ensure hemostasis.

Before the sutures are placed to further correct ear prominence, pressure on the helical rim and antihelix again demonstrate that the spring or resistance of the cartilage to further antihelix shaping is significantly decreased, and the reshaping can be accomplished without any sharpening of the antihelix. This is a key point of this otoplasty technique, because even a limited amount of conchal resection will result in this lessened spring and increased ease of shaping of the antihelix. With the current exposure of the posterior surface of the concha, additional sutures can

be placed before the key posterior shaping sutures if they are needed to complete an accurate closure of the conchal defect.

Two or three 4-0 clear nylon sutures are placed from scaphal cartilage and helical sulcus cartilage to mastoid fascia to correct the prominence of the upper and middle thirds of the ear. The third, most superior, suture may be placed from the region of the triangular fossa to the temporal fascia. The proper placement of these permanent sutures is determined by applying pressure to the desired points on the anterior surface of the ear with a straight Keith needle, then passing the point of the needle through the selected spot so it can be viewed on the posterior surface of the cartilage (see Fig. 32-3, *I* and *J*). The nylon suture is then placed at each specific point without needing to tattoo the cartilage. As the suture is placed, the Keith needle is withdrawn. The sutures are then passed through mastoid or temporal fascia. Once all of the sutures are placed, they are tied, beginning with the scaphal suture, to the proper tension to create a smooth antihelical fold. Before cutting each suture, a forceps is used to grasp the knot and slide it down against the mastoid surface, minimizing any risk of later extrusion of sutures. As these sutures are tied, care should be taken to observe the relationship of the reshaped antihelix and the helical rim to ensure that the helical rim remains visible from the frontal view. The suture from the triangular fossa to the temporal fascia creates a more natural line from the upper pole of the ear to the helical root. Again, the exact suture position and closing tension is easy to control, and subtle differences can be easily observed.

Once the upper and middle auricular prominences are corrected, the lobule position is addressed by closing the posterior squid-shaped defect with 5-0 chromic gut. With the diamond-shaped inferior portion of the squid design previously described, the appropriate contour, shape, and projection of the lobule are created by varying the initial suture position, with *A'* (the point of the diamond at the point of maximal lobule prominence) being sutured to *A*, *B*, or *C* (more frequently to *A* rather than to *B* or *C*) (see Fig. 32-3, *K* and *L*). Once this initial suture is placed, the remaining closure is accomplished with a running 5-0 chromic suture. Correction of the more prominent side first allows for a symmetrical “setback” by providing a guide for placing the sutures that define the antihelix and lobule prominence (Figs. 32-4 and 32-5). Finally, as one becomes more comfortable directly approaching minor cartilage irregularities or areas of residual prominence, such as an antitragus or a Darwin’s tubercle, one can directly resect the area of cartilage prominence through a small additional incision along the border of the area of cartilage to be trimmed and repair the skin with 6-0 chromic suture. These additional incisions are virtually invisible when healed, and this type of approach is very much a part of correcting many of the other nonmicrotia deformities.

Macrotia

The approach to prominent ears, as previously discussed, must be flexible and adjusted to the unique features of each deformity. Cases of true macrotia require reduction of the overall ear size in addition to correcting ear prominence. In general, reducing the vertical height of the ear is most easily accomplished by using an excision at the junction of the middle and lower thirds of the ear in conjunction with a helical rim advancement using a posteriorly based chondrocutaneous flap. The excess cartilage is excised from the scaphal region, the rim is advanced along, and additional excess skin and cartilage are excised just inferior to the helical tail. This approach affords an excellent view for either further development of the antihelical fold or additional conchal reduction.



Fig. 32-4 A-D, Preoperative views of a 7-year-old girl with prominent ears caused by mild conchal hypertrophy, in combination with asymmetrical effacement of the upper antihelical folds. E-H, Results 6 months after the combined otoplasty approach was used. Note the excellent correction with smooth antihelical folds and the natural visibility of the helical rim in the anterior view.

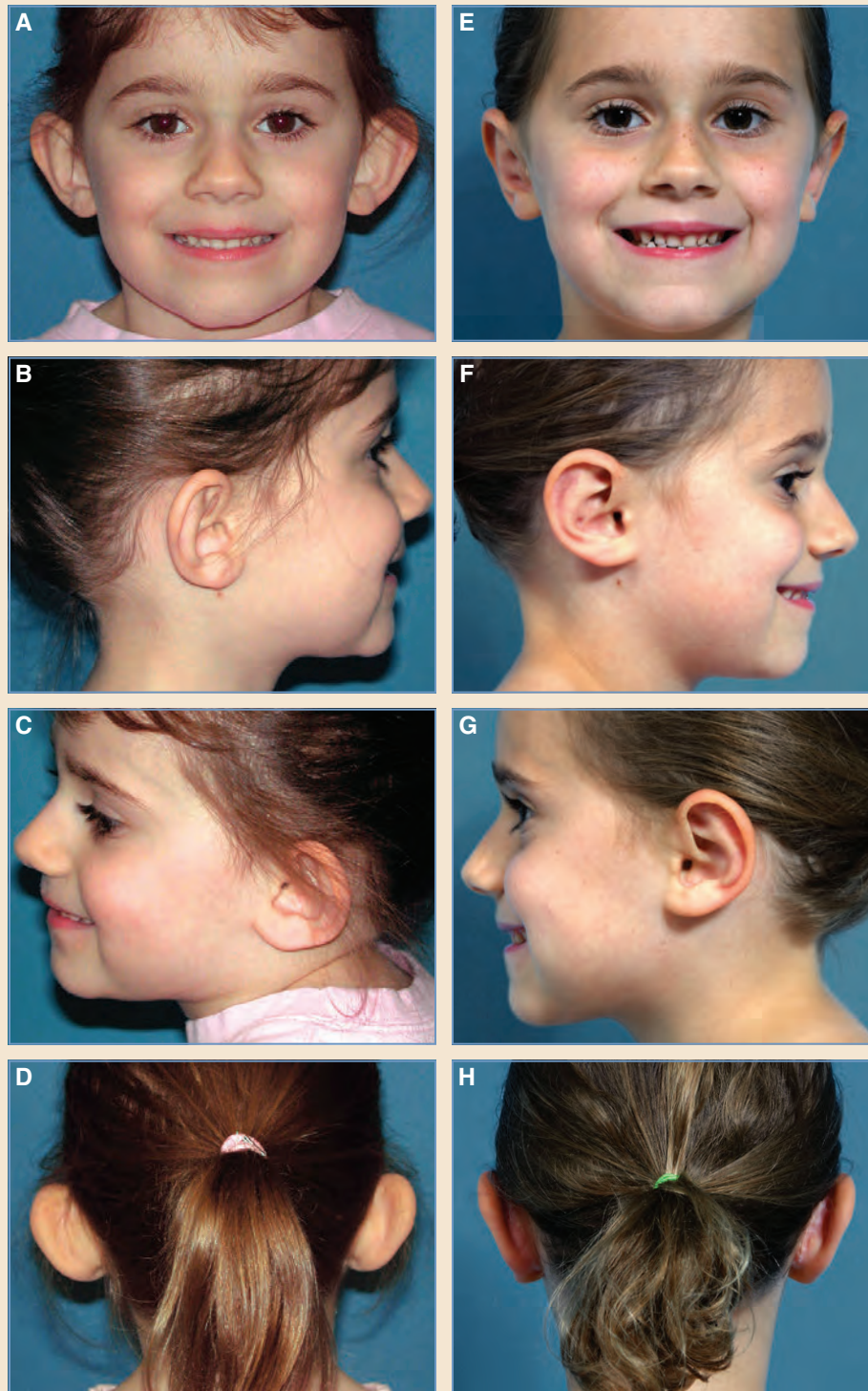


Fig. 32-5 A-D, Preoperative views of a 5-year-old girl with prominent ears caused by conchal hypertrophy and mild effacement of the antihelical folds (left to right). E-H, Results at 6 months postoperatively. Note the lack of distortion of the postauricular sulcus.

Constricted Ears

Anatomy

A constricted ear was previously described in the literature using a broad range of confusing terms such as *cup ear* and *lop ear*. The terminology for this group of congenital ear deformities was simplified by Tanzer,²³ who classified this entire group of anomalies as *constricted ear*. Tanzer²³ further divided constricted ear types based on both the degree of cartilage deformity and the extent of skin deficiency. Group I deformities are minor, involving the helix alone and giving a lidded appearance to the ear. Group II deformities are moderate to severe, involving the helix and scapha, and are further subdivided into group IIA without skin deficiency and group IIB, which requires supplementation of skin at the margin of the auricle. Group III deformities have extreme cupping with a tubular form to the ear, with great similarity to a large chondral remnant type of microtic deformity (Fig. 32-6). Group III deformities are often associated with deformities of the external auditory canal and middle ear.

Cosman²⁴ described four fundamental aspects of the constricted ear deformity that vary depending on its severity: (1) lidding, (2) protrusion, (3) decreased ear size, and (4) low ear position. These features correlate quite well with Tanzer's groupings.²³ *Lidding* is caused by helical overhang, arch shortening, and flattening of the antihelical crura. It may be further accentuated by a crimping of the rim with adhesion to the scapha. Decreased ear size is the result of the previously described changes in addition to a decreased skin envelope, conchal widening and angulation, and an actual decrease in the overall size of the cartilage. A low ear position is not unlike that seen in a variety of severe auricular malformations and is sometimes related to other associated skeletal deformities and syndrome-related anomalies.

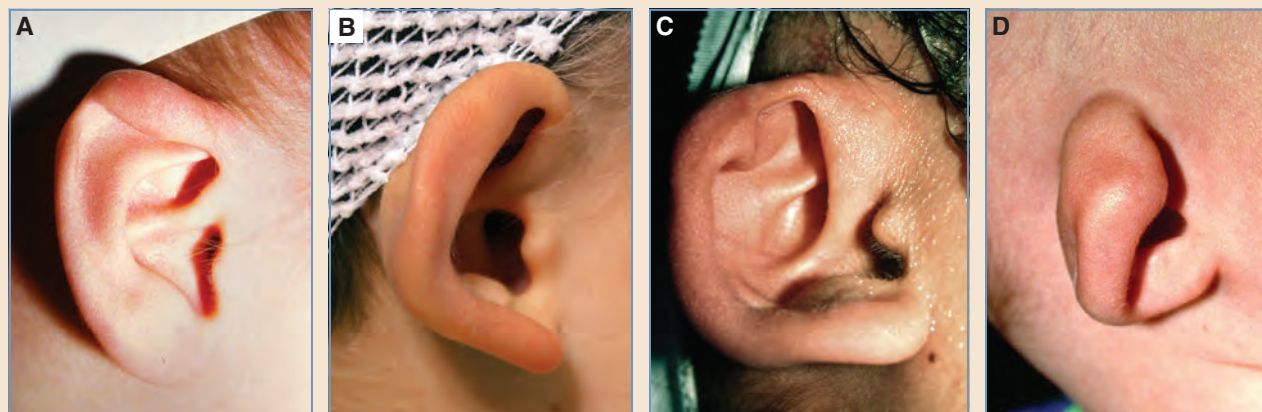


Fig. 32-6 Constricted ear deformities are grouped on the basis of the degree of lidding, the decreased ear size as the radius of the curve of the helical rim decreases, and the degree of skin shortage. **A**, Group I, with lidding alone. **B**, Group IIA, with combined lidding and increased constriction of the helical rim, but without significant skin shortage. **C**, Group IIB, with greater constriction and skin shortage. **D**, Group III, with skin and cartilage deficiency comparable to a large conchal remnant microtia.

Treatment Guidelines for the Constricted Ear

Because the presentation and spectrum of constricted ears are so variable, the treatment must be individualized to the specific deformity. Regardless of the technique chosen, the specific aims of treatment are to establish maximum (or near-normal) vertical height to the ear and to correct the ear prominence.²⁵ Techniques vary in their ability to increase the vertical height through cartilage reshaping alone, or in combination with additional contralateral auricular cartilage grafts or a costal cartilage graft. Before looking at the approach that we now use, which is equally applicable to most of the other nonmicrotia deformities (constricted ear, cryptotia, Stahl's ear, and a group of atypical unnamed deformities), it is worthwhile reviewing the techniques historically applied to the constricted ear.

One consistent shortfall for the majority of techniques described in the literature is that few techniques are capable of establishing a normal vertical ear dimension.^{23,25,26} In fact, a careful review of the 17 techniques illustrated in Cosman's excellent paper on the constricted ear²⁴ reveals that only one technique accomplishes this normal vertical ear dimension. In cases of bilateral constriction, obtaining symmetry between the two sides may not be a problem, but when the contralateral ear is of normal size, one must either accept that there will be a discrepancy between the two sides, or as suggested by Brent,¹² the size of the normal ear must be reduced to match the reconstructed side.

With this in mind, the key elements in correction of the constricted ear are:

1. Placement of the incision along the line of the future corrected helical rim
2. Full visualization of the constricted cartilage of the upper half of the ear
3. Release of all adhesions in the cartilage
4. Expansion of the cartilage with conchal cartilage grafts
5. Redraping of the anterior skin flap over the reshaped cartilage of the upper pole of the ear

With the incision moved to an anterior position, the skin envelope can redrape in a way that was not possible with the previously accepted posterior incision, and allows the skin to accommodate a considerable increase in the size of the upper pole. Although early experience with this approach will raise concerns about the ability to accomplish this "expansion" of the ear while maintaining a healthy vascular supply to the anterior skin flap, meticulous dissection of the cartilage in the subperichondrial plane will leave a healthy vascular flap. The redraping is further assisted by multiple fine quilting sutures to see that there is no tenting of the skin as it redrapes. These principles follow through all cases of constricted ear, although in varying degrees, as we will see when looking at each of the deformities.

Group I Constriction

Treatment of group I constriction is directed at two elements: the lid of extra skin and cartilage and the prominence of the upper pole. In fact, it is not uncommon to find an element of this type of constriction with many of the more typical prominent ears. More recent experience has demonstrated that in both group I and group II constrictions, there is often a degree of crimping of the helical rim (in addition to the lid) where there is an acute angulation between the helical rim and scaphal cartilage. Often the flattened rim is adherent to the scapha, with an apparent deficiency of skin in the area of adherence. Treatment is similar with either presentation and the maneuvers required will apply equally well to the crimping and distortion of cartilage in the upper pole in cases of cryptotia and Stahl's ear.

In our experience, it is rare that trimming of the lid alone will give an optimal result. Even with the release of any fibrous adhesions, the superior helical rim may still not unfold to its maximum height, and without support of the released cartilage with a “batten” of cartilage graft, it will still tend to droop. Once the graft supports the upper pole, the skin is redraped using multiple 6-0 chromic sutures to “quilt” the skin into the contours of the reshaped cartilage and prevent tenting of the skin, and the helical rim incision is closed.

In the procedure illustrated, the lid or hood of extra tissue is outlined along the line of the planned helical rim (Fig. 32-7, *A* and *B*). The excess tissue is trimmed and the rim repaired (Fig. 32-7, *C* and *D*). If the upper pole is also prominent, an ellipse of skin can be excised from the postauricular surface in the upper sulcus, and a suture or sutures (4-0 clear nylon) can be placed from the scaphal area to the mastoid and tied to correct this prominence. If there is concern about

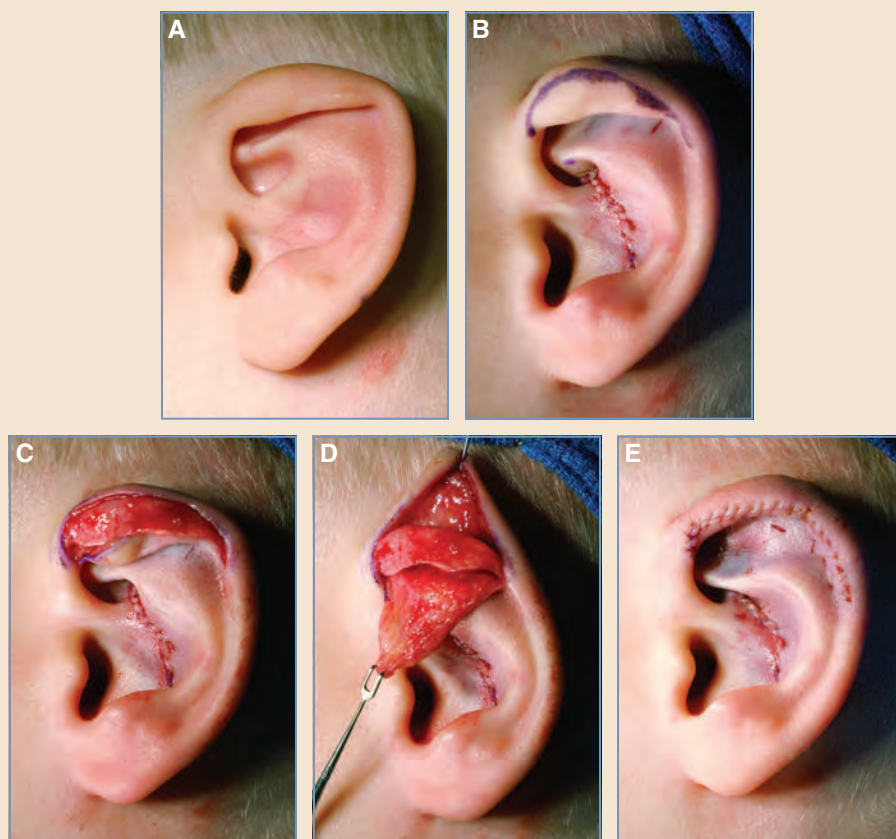


Fig. 32-7 Correction of lidding in group I deformity. **A**, The deformity. **B**, The incision is planned along the proposed new border of the helical rim, and the skin flap of the lid is dissected free to view the excess cartilage, which is trimmed. **C**, The skin flap is redraped, and the excess is excised. (As shown, the flap is split first, then the excess is trimmed after placing the central suture.) **D**, The helical rim incision is closed. Also shown is the conchal incision that was used to correct the associated ear prominence. **E**, At the completion of the procedure, the incision now lies along the border of the helix. Multiple quilting sutures can be used to ensure that the skin redrapes without tenting.

the vascular supply to the intervening flap that arises with a wide posterior skin excision, an alternative approach is to place a single incision at the auriculocephalic angle and tunnel the suture down from the anterior incision through to the sulcus. The scaphomastoid sutures can then be passed through these incisions without concern for the vascular supply. The postauricular skin closure is carried out in a similar fashion to that described for prominent ears, and the helical rim is repaired using 6-0 mild (fast-absorbing) chromic sutures (Fig. 32-7, E).

Group II Constriction

Although historically group II constricted ears have been separated into groups IIA and IIB based on the degree of skin deficiency, in all practicality, all group II deformities have some degree of skin deficiency. It has been our experience that the correction of group II constrictions, with increasing degrees of constriction of the skin and cartilage, presents perhaps the greatest challenge in ear reconstruction surgery. At the point when they are better classified as large conchal remnant microtia,²⁷ the approach is simplified through an expected staged reconstruction with lateral augmentation of the skin envelope in second-stage reconstruction. Although degloving the ear and expanding the cartilage with banner flaps²³ or upward and outward advancement of the cartilage of the cavum concha carried on the helical crus is capable of unrolling the constriction,²⁵ the newly enlarged ear frequently lacks strong vertical cartilaginous support. Once the skin is redraped over the cartilage, some of the height is lost. The addition of a conchal cartilage graft (ipsilateral, contralateral, or both) is frequently necessary to improve and strengthen the vertical correction; however, without the addition of skin, usually in the middle third of the ear, even the extra skeletal support may be inadequate. The cartilage harvest in each is readily accomplished as part of correcting the inherent prominence of these deformities as well.

Again, years of experience have led to the realization that degloving the upper pole of the ear beginning with a posterior incision may not only increase the risk of vascular compromise when the skin is dissected extensively onto the anterior suture, but also and more importantly, the redraping of the skin is more limited, because the constricted skin cannot fully splay out over the new framework. Approaching the deformity through an incision along the planned new helical rim allows the anterior skin to better adapt to the new shape without tethering and pulling down on the new upper pole framework. This approach allows one to gain significantly great vertical height without the need for additional posterior skin flaps. For symmetrical group IIA deformities, the upper pole corrections can be used as an adjunct to the previously described combined otoplasty techniques with excellent correction (Fig. 32-8).

For the benefit of comparing varied approaches, we have in the past successfully reconstructed some group IIB deformities with the addition of posterior skin flaps. The technique illustrated in Fig. 32-9 is similar to one described by Kislov²⁶ in that the constricted ear is split in its middle third, allowing the ear to open to a normal vertical dimension with a postauricular flap to cover the defect. A crescent-shaped chondrocutaneous flap from the concha (see Fig. 32-9, C and D) aids in the correction of the ear prominence and provides the needed tissue.²⁸ However, the Kislov approach²⁶ and variations of it have the downside of leaving transverse-oriented scars in the lower antihelix where the flap is spliced into place, and the result still does not have the flexibility achieved with the additional skin and cartilage added to the constricted upper pole of the ear that can be gained with altered placement of the incision, upper pole cartilage restructuring and a graft, and redraping of the skin (Figs. 32-10 and 32-11). If the deformity is unilateral and additional cartilage is needed, it can be harvested from both the ipsilateral and contralateral ears, but in most cases of bilateral constriction, the technique shown in Figs. 32-10 and 32-11 can yield near-normal height and superior shape.

Text continued on p. 964



Fig. 32-8 Correction of group IIA constriction in a patient with symmetrical constriction bilaterally. **A-D**, Preoperative views show ear prominence, decreased ear size, and lidding. **E-H**, Results 1½ years postoperatively. Correction was accomplished using a combination of maneuvers to correct lidding and the previously described combined otoplasty technique.

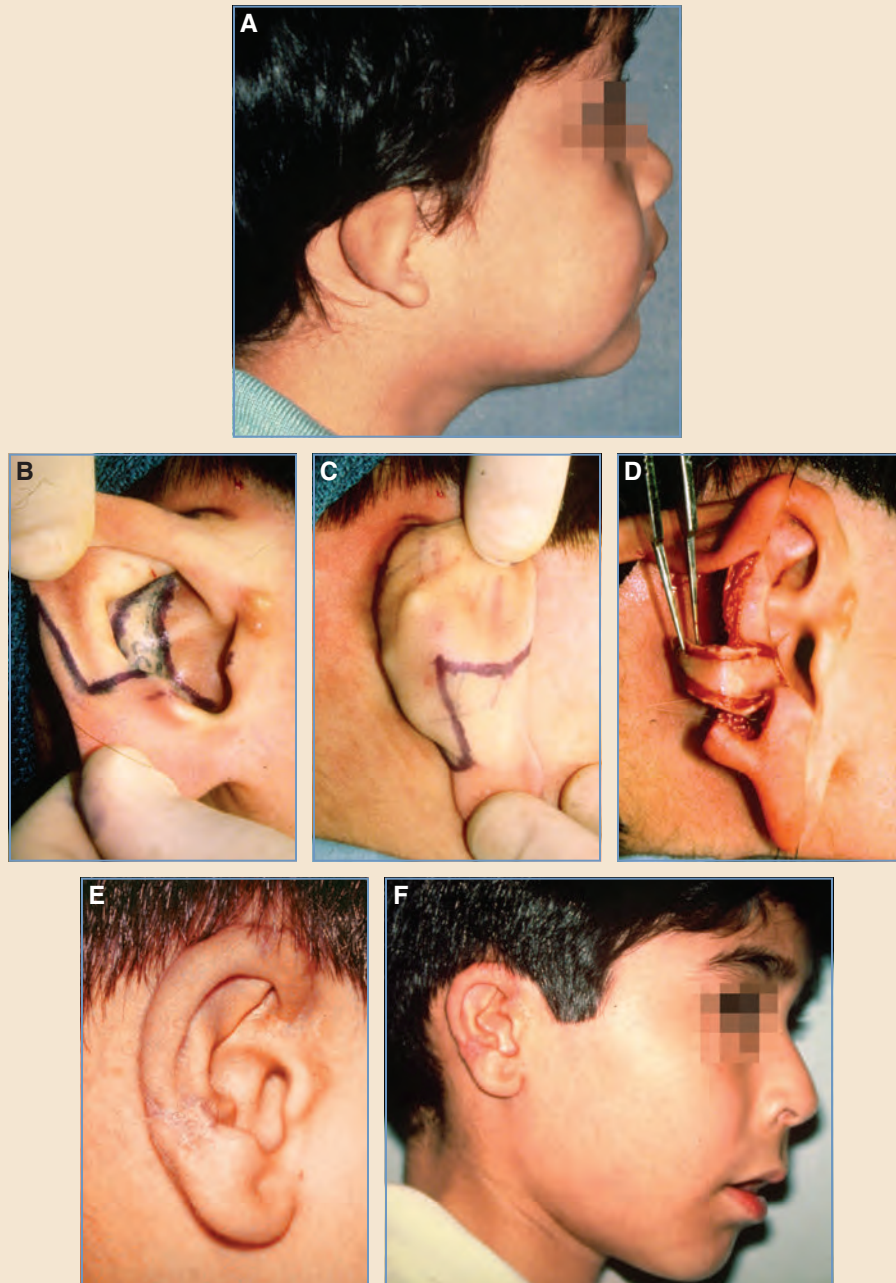


Fig. 32-9 Correction of group IIB constriction with flaps similar to the technique described by Kislov, but using a chondrocutaneous flap rather than a graft to fill the defect. **A**, Preoperative view. **B** and **C**, The planned incisions are marked. **D**, A chondrocutaneous flap is fitted into the defect created by splitting the constricted rim. Also shown is the postauricular skin flap that will wrap forward over the helical rim to join the edge of the chondrocutaneous flap. **E** and **F**, The corrected constriction approximately 1 year postoperatively.



Fig. 32-10 Correction of group IIB constriction. **A-C**, This child, seen at surgery, had asymmetrical constriction with the more significant deformity of the left ear, which shows a constricted scapha, lid-ding, prominence, and skin deficiency. **D** and **E**, The upper pole of the cartilage was approached anteriorly through an incision along the proposed future helical rim and the constricted, fully exposed cartilage. **F-H**, The cartilage lid was trimmed after exposure and set aside as additional graft material, and the upper pole was released or split in a manner to provide the best contour to the upper pole. In each case, the scapha was released with incisions planned to allow the most stable repositioning and fixation of the segments to the graft. Here it was split transversely until the constricted upper pole opened out.

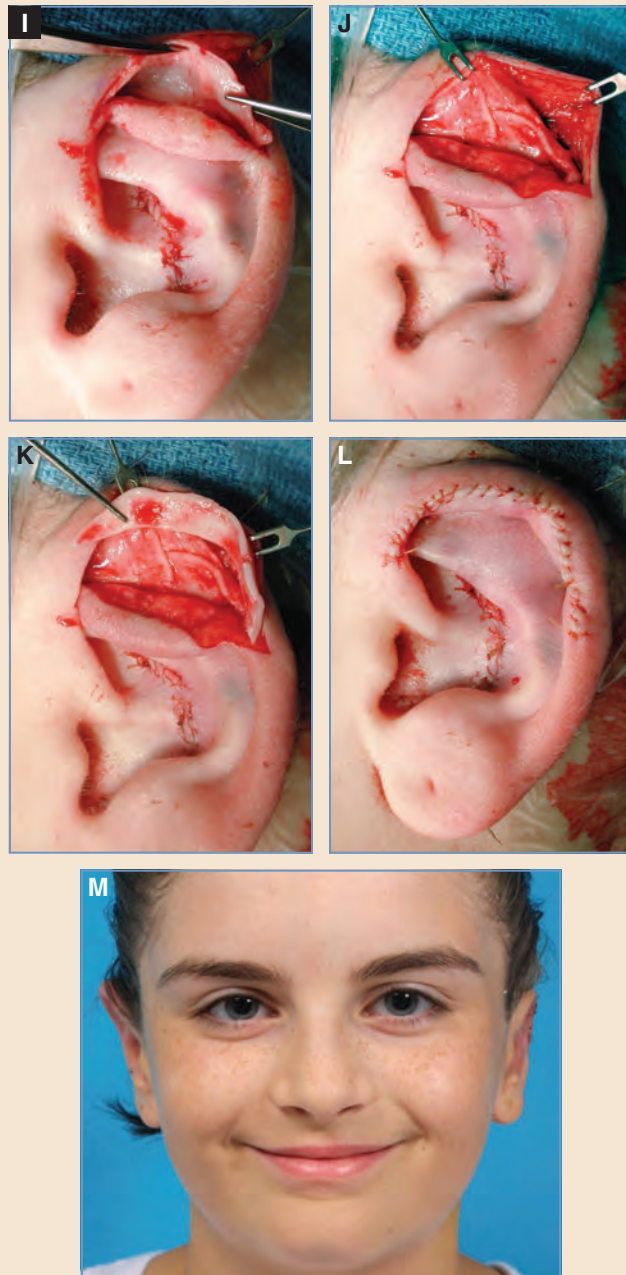


Fig. 32-10, cont'd I-L, The cut segments of the scapha were overlapped or spliced together and combined with the conchal cartilage graft. The resected segment of the helical rim lid was used to fill the residual rim defect after first setting the projection of the helical rim lid with supporting scapha to the mastoid and triangular fossa to the temporal fascial sutures. The skin was redraped with multiple quilting sutures, ensuring that there was no tenting of the skin over the cartilage contours, and the helical rim incision was closed. M, The correction is seen 2 weeks postoperatively.

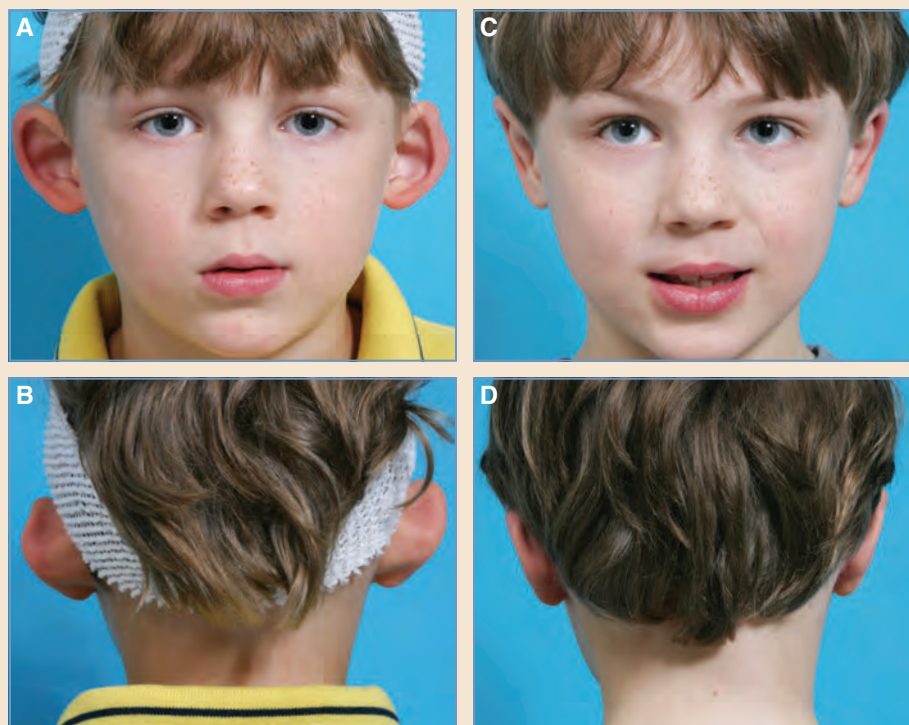


Fig. 32-11 Correction of asymmetrical deformities with prominence, constriction, and acute crimping of the helical rim using a similar approach to that in Fig. 32-10. The patient is seen preoperatively (A and B) and 3 months postoperatively (C and D).

The grafts to the upper pole of the ear are spliced into place with 5-0 and 6-0 clear nylon sutures, placing the sutures in such a manner as to secure the adjacent overlapping cartilage edges for a smooth transition. Once the primary grafts are in place and the greater part of the grafting is completed, some supporting sutures of 4-0 clear nylon are placed in a similar fashion to those used in the typical correction of the prominent ear. These sutures set the projection of the upper pole (sutures from scapha, concha, and triangular fossa to the mastoid and temporal fascia). Once this is accomplished, the final trimming of the helical rim or an additional graft can be placed. (Typically a remnant of the original lid may be needed at the junction of the upper pole and midhelical rim for a smooth transition when seen in frontal view.) Not uncommonly, in bilateral cases in which there is limited available additional conchal cartilage, the thin rim of the original lid is available for this final graft.

An additional point that is pertinent to the treatment of constricted ears, as well as the remaining upper pole deformities, relates to the harvest of cartilage. In each of these cases, the harvest of the conchal graft aids in correction of the ear prominence associated with many of these deformities. Depending on how much graft is harvested, the conchal cartilage edges may not be approximated, and there will be a conchal defect, as in any case where a large conchal graft

has been harvested in other reconstructive procedures. The cartilage defect will not be visible if the skin is closed without tension and the overall shape of the antihelix has not been disrupted.

Group III Constriction

In most cases the treatment for group III constriction is the same as the treatment for the so-called *conchal-type microtic remnant*.²⁷ There is a severe deficiency of both cartilage and skin, and reconstruction typically requires placing a costal cartilage framework, which is spliced into the usable parts of the constricted cartilage remnant. Following the skin flap design of Firmin²⁹ may provide the best use of the skin flaps. The reconstruction, similar to that for microtia, generally requires two or three surgical procedures to fully splice the parts together and complete the elevation of the helical rim and create the postauricular sulcus. With increased experience with the techniques described for the treatment of group IIB constrictions, unilateral cases of group III constrictions with a large conchal remnant may be well reconstructed using combined ipsilateral and bilateral grafts and auricular flaps without the need for costal cartilage. With this approach, the final result will have finer cartilage detail and a more naturally flexible ear, but the height may be slightly shorter than that gained with placement of a costal cartilage graft. The reconstruction may also require a second surgery.

CRYPTOTIA AND STAHL'S EAR

A number of congenital ear deformities, in addition to those described previously, involve varied deformation of the antihelix and upper pole of the auricle. Although these deformities are not common in whites or other races in the United States, they are seen with fairly high frequency in the Asian population (cryptotia occurs in approximately 1 in 400 to 500 Asian births).³⁰ Because these deformities are relatively rare, most surgeons have limited experience treating them, and although an in-depth discussion of each is beyond the scope of this chapter, I think it is worthwhile to review their anatomy and make several key points concerning their evaluation and treatment. The key point to understand in the correction of congenital ear deformities involving varied deformation of the antihelix and upper pole of the auricle, as well as the constricted ear deformities previously discussed, is that it would be much better to put many of the classifications and names aside and consider this entire group of ear deformities, and many other atypical unnamed defects, to be upper pole deformities. Each of these deformities has an element of deformation or deficiency of cartilage, in part because of fibrous adhesions, and a varied degree of skin shortage. Using the approach described earlier to visualize the cartilage and address the cartilage deformity, the reconstructions can be readily accomplished.

Cryptotia: Anatomy and Treatment

Cryptotia is characterized by failure of the upper pole of the ear to stand out from the head (Fig. 32-12). The three basic features of the deformity were described by Washio.³⁰ I have added a fourth feature that may need to be addressed for optimal outcome:

1. Buried superior pole of the auricle beneath the scalp skin
2. Scaphal underdevelopment
3. Sharpening of the antihelical crura, particularly the superior crus
4. Helical rim crimping and adhesion

Hirose et al³¹ thought that this deformity was the result of an anomaly of the intrinsic transverse and oblique auricular muscles. Based on this theory, these investigators divided cryptotia into type I (transverse muscle type), in which the body of the antihelix and the superior crus are compressed together, and type II (oblique muscle type), in which there is gross contraction of the body of the antihelix and an acutely bent inferior crus.

Treating cryptotia, regardless of the technique chosen, consists of releasing adhesions, reshaping cartilage, and creating the superior helical sulcus with a skin flap closure. According to Washio,³⁰ the most important element of the correction is the release of all adhesions on the posterior aspect of the antihelix (see Fig. 32-12, *E*), combined with reshaping the superior crus using suture fixation. Once the upper pole of the ear is projected away from the scalp, the posterior defect can be repaired using the basic principles of plastic surgery. Rotation flaps and V-Y advancement flaps have been used most commonly, advancing skin from inferiorly and posteriorly in the sulcus into the superior sulcus.³⁰⁻³² If the deformity includes a crimping and adhesion of the superior helical rim, this deformity should be addressed as well by releasing anterior adhesions and splinting the newly shaped helical rim with a batten graft of conchal cartilage (see Fig. 32-12, *F*). Although the greater part of the deformity can be completed without addressing the anterior crimp, and previous descriptions of the treatment of cryptotia have not included this added correction, the final outcome is more refined when the anterior deformity is also addressed.

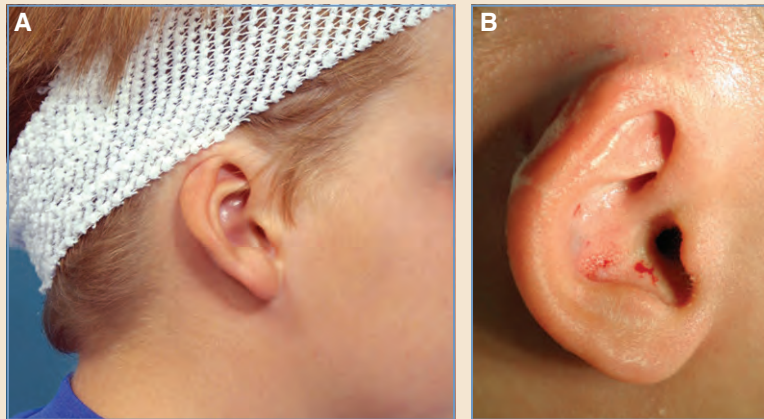


Fig. 32-12 Anatomy and correction of cryptotia. **A** and **B**, Typical deformity of cryptotia shows the buried upper pole of the ear and distortion of the upper pole. The deformity is most commonly caused by acute folding of the superior crus resulting from posterior adhesions, and may also include crimping and distortion of the helical rim as a result of anterior adhesions.

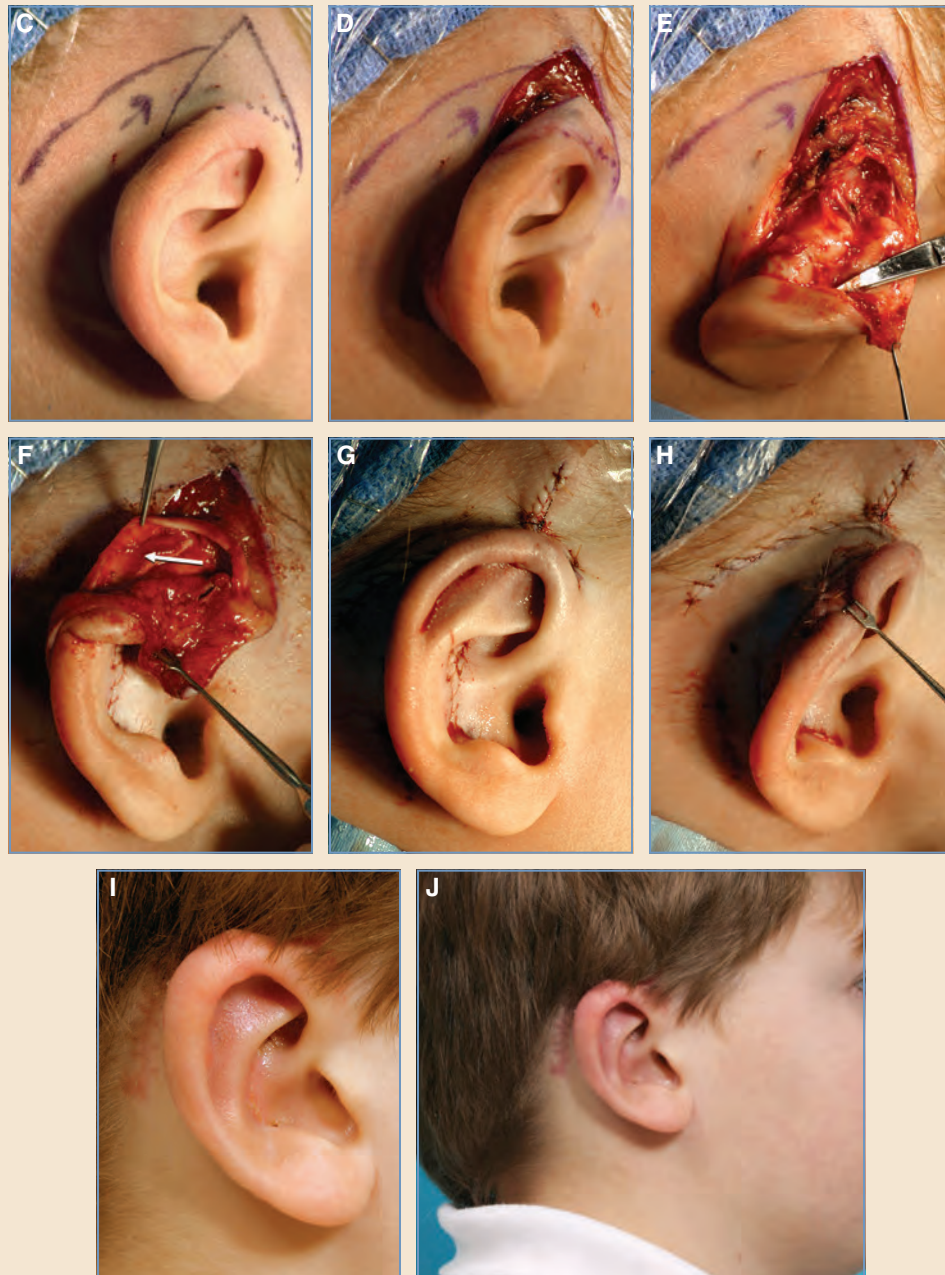


Fig. 32-12, cont'd **C**, Incisions are designed to develop flaps to cover the superior helical rim and advance into the defect to create the superior sulcus. **D**, The superior pole of the ear is released from the scalp. **E**, All adhesions are released from the postauricular surface of the superior crus. **F**, The skin flap has been elevated to expose the anterior surface of the helical rim and scapha, allowing the release of adhesions and placement of a batten of cartilage (*arrow*) to splint the rim in its corrected position. **G** and **H**, The anterior flap is redraped over the corrected cartilage, tacking the skin flap into the cartilage with through-and-through fine chromic sutures, and the postauricular flap is advanced into the defect to line the sulcus. **I** and **J**, One month after correction, seen in close-up and wider views, showing the natural shape of the upper pole and the well-defined superior sulcus.

Stahl's Ear: Anatomy and Treatment

Stahl's ear, an unusual deformity that is sometimes referred to as *Spock's ear*, has as its main feature a third crus in combination with a flat helix and malformed scapha.³³ The third crus typically projects from the antihelix at almost a right angle outward toward the helical rim, and in some cases there is only a displaced superior crus and not three crura. This deformity forces the helical rim up into a point at the junction of the middle and upper poles of the auricle (Fig. 32-13, *A*). I think that there is also a group of congenital ear deformities in which there is just a vertical lengthening of the superior crus (without a third crus) that creates a similar distortion of the helical rim and pointing of the ear, and I consider this to be a variant of Stahl's ear (Fig. 32-13, *B*).

Historically, treatment of Stahl's ear has consisted of direct exposure of the malformed cartilage, cartilage Z-plasty or reversal, and cartilage suture techniques.³³⁻³⁵ However, none of these techniques has been consistently effective in fully correcting the deformity. Applying the approach described for the whole group of upper pole deformities, with direct visualization of the deformed cartilage, these deformities will effectively be corrected through direct release of the deformed cartilage, use of conchal cartilage graft, appropriately placed sutures to reshape the antihelix, and redraping of the skin (Figs. 32-14 and 32-15), as well as a whole host of deformities that defy specific description beyond noting the specific anatomic defects.

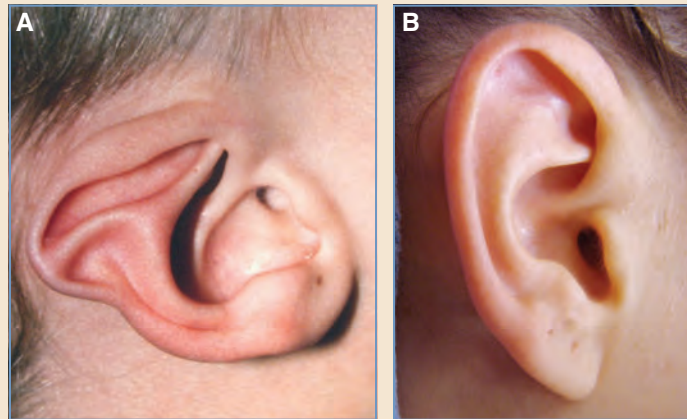


Fig. 32-13 *A*, Stahl's ear deformity has as its main feature a third crus in combination with a flat helix and malformed scapha. *B*, A group of deformities exists in which the upper pole is elongated by a long superior crus and elongated scapha that results in a pointed ear. I consider this group of deformities to be variants of Stahl's ear.



Fig. 32-14 Stahl's ear corrected with reshaping the upper pole through the direct approach and conchal cartilage graft in addition to correction of ear prominence on the contralateral side. **A-D**, Anterior and lateral views in wide and close-up preoperative images show the addition of crimping and adherence of the helical rim and third crus. The left ear is prominent. **E and F**, The patient is seen 10 years after correction of the prominence and helical rim deformity. The procedure involved the exposure of upper pole cartilage, release of adhesions, and conchal cartilage graft to support the reshaped helical rim.

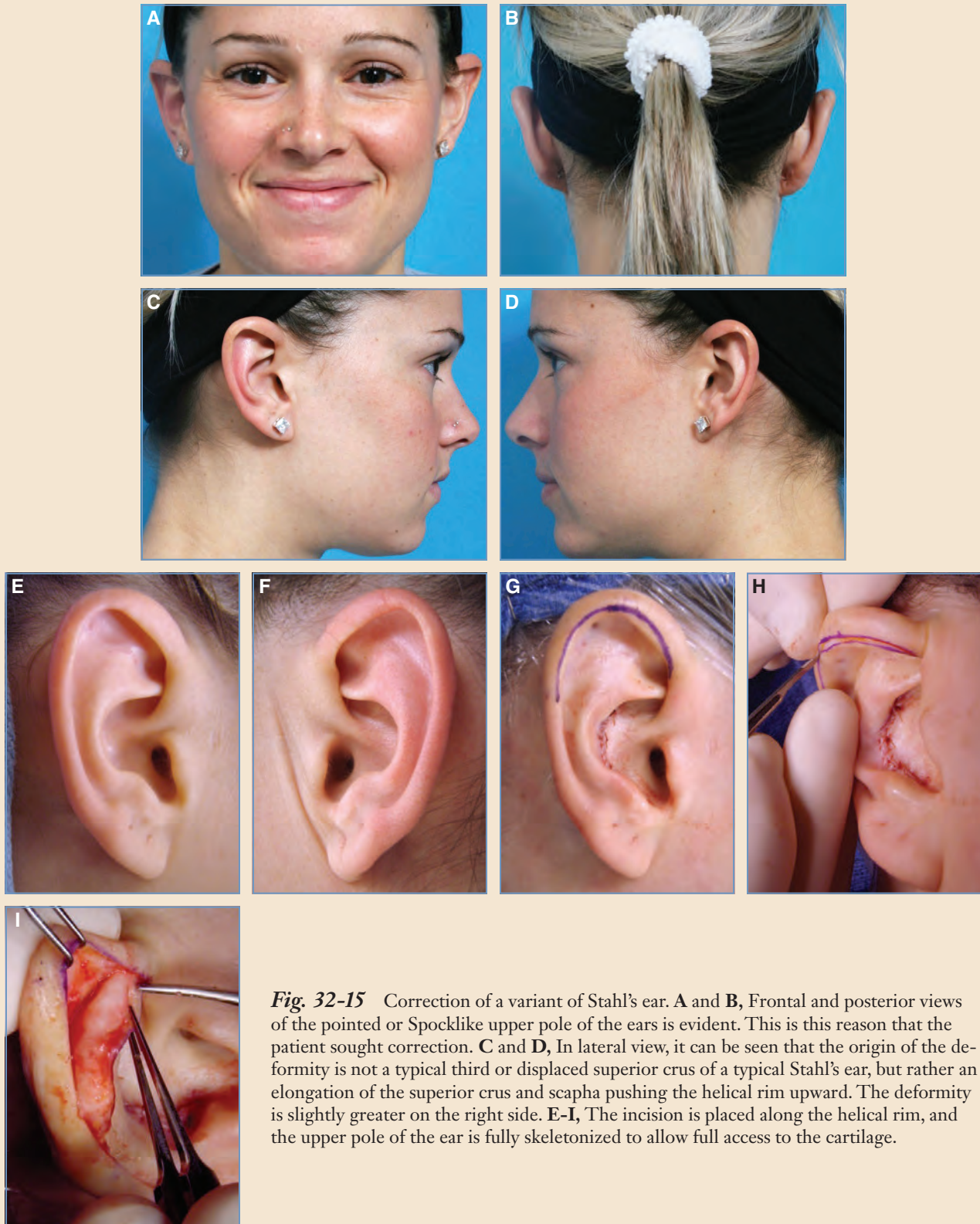


Fig. 32-15 Correction of a variant of Stahl's ear. **A** and **B**, Frontal and posterior views of the pointed or Spocklike upper pole of the ears is evident. This is this reason that the patient sought correction. **C** and **D**, In lateral view, it can be seen that the origin of the deformity is not a typical third or displaced superior crus of a typical Stahl's ear, but rather an elongation of the superior crus and scapha pushing the helical rim upward. The deformity is slightly greater on the right side. **E-I**, The incision is placed along the helical rim, and the upper pole of the ear is fully skeletonized to allow full access to the cartilage.

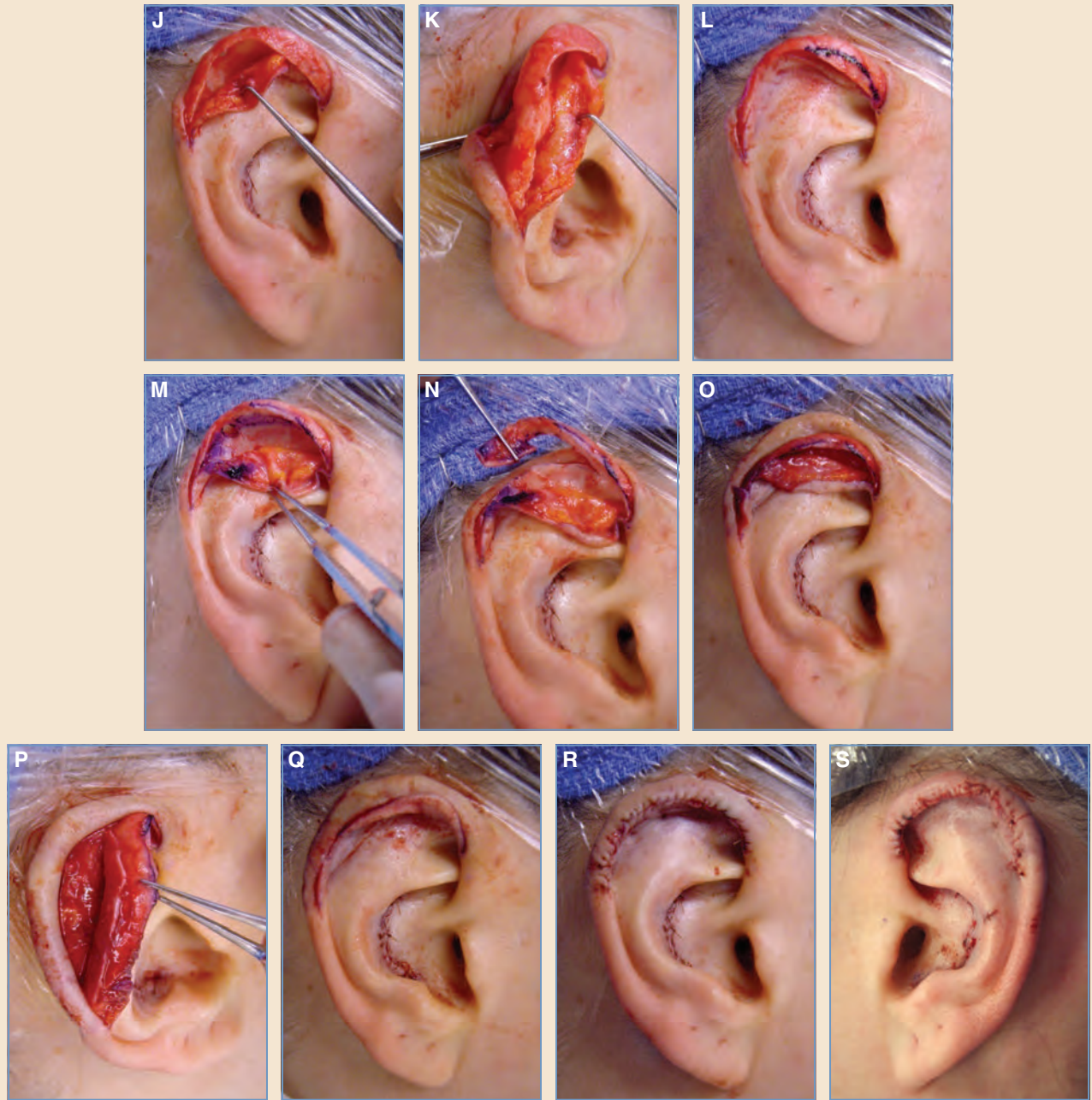
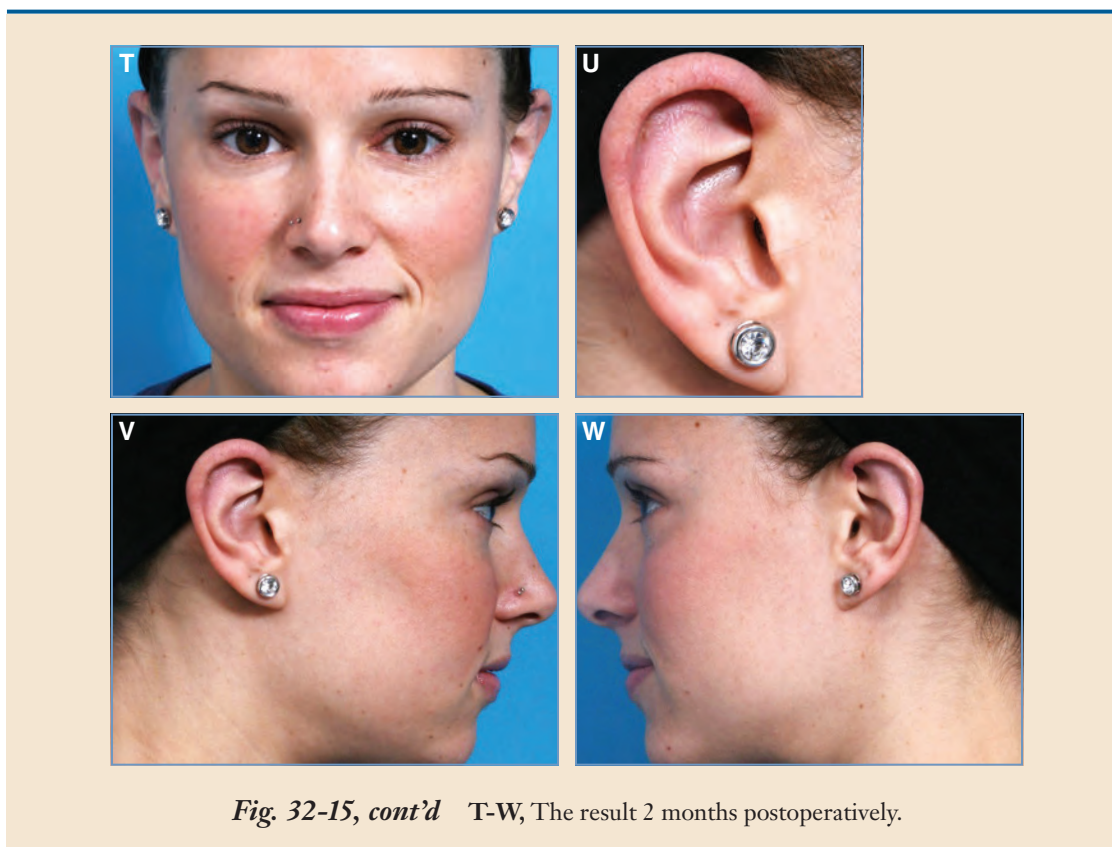


Fig. 32-15, cont'd **J**, The anterior lid of extra cartilage is outlined and resected. **K-N**, The helical rim cartilage is freed and a portion resected, and then the shortened helical rim is fixed in place with sutures to the junction with the midthird of the helical rim and to the shortened scapha. **O**, The skin is redraped and the excess resected. **P** and **Q**, The completed correction is shown in comparison with the elongated preoperative shape; the ear has been shortened and has a more natural curve to the upper pole. **R** and **S**, Close-up views of the right and left ears after correction.

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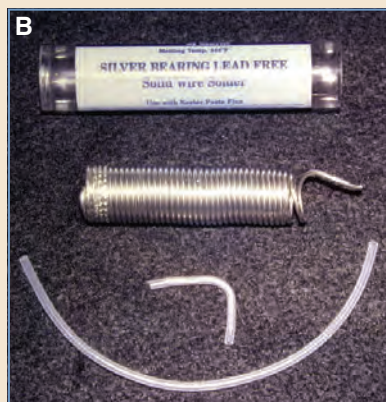
NONOPERATIVE TREATMENT OF CONGENITAL EAR DEFORMITIES

In addition to the surgical techniques previously described for the treatment of prominent ears, constricted ears, cryptotia, and Stahl's ear, each of these deformities may be treated and corrected at least partially by nonoperative splinting techniques. First described by Matsuo et al³⁶ and then by Brown et al,³⁷ the nonoperative approach is based on experimental evidence that auricular cartilage is soft and malleable during the neonatal period while maternal estrogen levels are still high. Once these levels drop (usually after 2 weeks of age), the cartilage becomes firmer and holds the shape into which it has been molded (Fig. 32-16).

This nonoperative approach has been applied most successfully to Stahl's ear, group I and II constrictions, and prominent ears when the primary feature is effacement of the antihelical fold. Although a variety of splinting materials and elaborate prefabricated splints have been used, I have found that lead-free solder with a covering silicone sleeve, taped on with Steri-Strips, is well tolerated and has never been associated with skin necrosis (a concern with harder splinting materials), and does not require the purchase of costly materials. Although Yotsuyanagi³⁸ has reported that splinting can be effective even in older children (6 years of age and older) with a thermoplastic splint applied for a relatively short period, this has not been substantiated by other investigators. It is my experience that effective nonoperative molding of the ear can be carried out until about 3 months of age. After that age, it is rare for the baby to leave the splint in place continuously, and rare for the parents to persist in replacing it. Deformities that do not involve significant skin shortage or sharp crimping of the cartilage and are associated with fibrous adhesions between cartilage surfaces are more amenable to nonoperative treatment.



Fig. 32-16 Nonoperative splinting in the neonatal period. **A**, A Stahl's ear deformity. **B**, The splinting material of lead-free solder and silicone tubing used for protective cover. **C**, The splint fitted to shape the ear from the helical sulcus to the triangular fossa. **D**, Another variety of rubber-coated wire with Steri-Strips holding the splint in place. **E**, The corrected deformity. **F** and **G**, A more complex deformity in a 2-week-old infant with features of both constriction and Stahl's ear. **H** and **I**, Six weeks after 5 weeks of splinting, with excellent correction.



When completing the splinting process, it is important for the parents to carefully observe the ear closely for 24 to 48 hours, because there may be some recurrence of the deformity that if monitored closely can be corrected through an additional week or two of splinting. In addition, it has been noted that of the varied deformities treated with nonoperative treatment, prominent ears are the least well corrected. This would seem to support the earlier discussion of the importance of the conchal size and projection being the underlying cause of most prominent ears. Splinting can quite effectively correct the effaced antihelical fold, but it has little or no benefit in correcting conchal shape.

CONCLUSION

The group of nonmicrotia ear deformities covers a broad spectrum of defects. Although some of these fall into well-recognized and named deformities, there are also many that defy naming and about which little has been written. If the deformities are separated based on the degree of prominence and whether or not the upper pole of the ear is deformed, it is possible to apply a systematic approach to their treatment that yields consistent natural results. The choice of otoplasty technique is not as important as picking the appropriate procedure for the specific deformity being treated. In addition, recognition that the otoplasty technique chosen must be tailored to the deformity, rather than the reverse, is critical to avoid an unfavorable result.

Chondrocutaneous conchal resection is the cornerstone of correction of the majority of prominent ear deformities, and it can also be the source of cartilage graft for reconstruction of upper pole deformities. A direct anterior approach to the upper ear, combined with the release of cartilage adhesions and “reshaping” of the cartilage, can be performed alone or in concert with the combined otoplasty techniques to predictably correct a spectrum of upper pole deformities. Understanding the common anatomic deformities, regardless of whether they fall under the classic description of constricted ear, cryptotia, or Stahl’s ear, is more pertinent to the successful correction of the deformity than is the name used to describe the defect.

KEY POINTS

- There is great variability among ears and among ear deformities, and each deformity must be assessed in relation to the normal ear anatomy of the patient and to the differences between the two sides.
- Ear position and projection are significantly affected by abnormalities in the underlying craniofacial skeleton.
- Although the previous literature has emphasized accentuation of the antihelical fold for correction of the prominent ear, my work has clearly demonstrated that chondrocutaneous conchal resection is the cornerstone of otoplasty.
- The group of upper pole ear deformities, including most constricted ears, cryptotia, Stahl’s ear, and some prominent ears (with crimping of the helical rim), can best be corrected with direct exposure of the cartilage defect through an anterior approach and release of cartilage adhesions.
- Whether correcting a constricted ear or an acquired ear deformity, a critical decision must be made: Will the reconstruction match the size of the opposite ear? Or should the emphasis be on normalizing the shape of the reconstructed ear and accepting a size difference?

- Recognizing that we rarely see both of an individual's ears at the same time, differences of 0.5 to 1.0 cm may not be very noticeable, and simpler procedures may often accomplish a better shape without matching vertical height.
- Nonoperative or neonatal splinting for correction of a variety of congenital ear deformities, of which Stahl's ear is the most readily corrected, can be accomplished with simple materials and is most effective if applied within an infant's first 3 months.

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33

Treatment of Major Acquired Ear Deformities

Bruce S. Bauer • Donald Brooks Johnson



The principles gained from treating major congenital ear deformities, especially microtia and the more severe forms of constricted ears, are directly translatable to treating major acquired ear deformities. Although for some surgeons acquired deformities are more a part of their practice than congenital deformities are, it is helpful to have a generalized method of classifying acquired deformities to be able to select the optimal reconstructive technique.¹⁻⁵ I (B.S.B.) have often thought that the simplest way to do this is to separate cases by both etiologic factors and the extent of the soft tissue deficiency. Etiologic factors can be classified as (1) trauma of disease or iatrogenic injury, (2) burn and post-chondritis deformities, and (3) traumatic (non-burn-related) auricular defects. Tissue deficiency can be classified as (1) skin and cartilage deficiency with minimal or “limited” areas of scarring (Fig. 33-1, *A* through *E*) and (2) skin and cartilage deficiency with moderate-to-severe scarring (Fig. 33-1, *F* through *H*).

A critical question that the surgeon must ask patients seeking reconstruction is whether the reconstruction needs to match the opposite normal ear in height, or whether the patient is willing to accept a well-formed reconstruction that may be smaller than the opposite ear? Because we rarely see both ears at the same time, differences of 0.5 to 1.0 cm may not be that noticeable, and simpler procedures may often produce a natural-shaped ear without matching vertical height.



Fig. 33-1 Acquired ear deformities can be classified according to whether there is limited or moderate-to-severe scarring and the extent of missing cartilage. This provides insight into the type of reconstruction that will be required. In **A-C**, three individuals with postchondritis deformity are shown here, each with a limited skin deficiency. **A**, This patient required a contralateral conchal cartilage graft alone. **B**, This individual had a combined ipsilateral and contralateral conchal cartilage graft. **C**, This patient had a costal cartilage framework. **D** and **E**, Two patients with traumatic ear loss are shown in which the adjacent skin is minimally scarred and could be elevated in continuity with the vestige for flap coverage of the replaced costal cartilage framework. **F**, A patient with a failed microtia reconstruction in which the skin is still supple enough to cover a new cartilage framework. **G**, Cartilage loss with moderate-to-severe scarring and loss of the greater part of the ear, with scarring of the surrounding skin resulting from an in utero vascular accident. **H**, This child had a failed reconstruction after eight attempts at reconstructing a lobular type of microtia.

Selection of a simpler approach may be warranted for older patients or when the deformity is the result of tumor resection when the long-term behavior of the resected tumor may be unpredictable. The decision may also be prompted by a desire to have the reconstruction completed with a single procedure rather than with staged procedures that might be required if the full vertical height were reconstructed.

It is possible to decide whether the defect can be reconstructed with adjacent skin flap coverage or whether a fascial flap and skin graft are needed for vascular coverage. It is also possible to determine whether the cartilage defect can be repaired with conchal cartilage (contralateral, ipsilateral, or both) or requires a costal cartilage graft.

An in-depth discussion of each of the many types of acquired deformities is beyond the scope of this chapter; however, a few basic guidelines can be added to the preceding that may aid surgeons in the selection of an ideal approach to the deformity. In general, if the quality of the skin is reasonably good (limited scarring of both the ear remnant and adjacent skin) and the defect is not too large, a more delicate reconstruction can be accomplished with conchal cartilage grafts rather than costal cartilage grafts. However, even if the defect can be bridged with conchal cartilage grafts, a costal cartilage graft may be preferable if fascial flap coverage is required, because conchal cartilage grafts are not sturdy enough to resist the contracting forces of the healing fascial flap. The vascular supply to the ear and postauricular area is excellent, allowing a multitude of flap designs. Although this vascular supply allows considerable ingenuity in choosing a procedure, careful planning can allow the use of the larger, more reliable flaps. Patients should understand that staged procedures are the rule rather than the exception, even though after the initial major reconstruction stage, subsequent procedures may be minor but necessary to obtain an optimal result.

SKIN AND CARTILAGE DEFICIENCY WITH LIMITED SCARRING

Soft tissue deformities with limited scarring, whether they arise from trauma, resection of a tumor, infection, or iatrogenic injury, are characterized by either significant loss of cartilage with minimal injury to the overlying skin (for example, postchondritis deformity) or loss of both skin and cartilage but with limited injury to the skin and surrounding vasculature.⁶ The postchondritis deformity is perhaps the best example of the acquired defect with limited scarring, although small bite injuries and segmental sharp avulsions may likewise fit into this category.

Postchondritis Deformity

The postchondritis deformity is an excellent example, because the skin envelope, even if contracted as a result of scarring from delayed resolution of the infection, is always sufficient to cover the new framework if it is delicately dissected from the remaining amorphous remnant (see Fig. 33-1). The choice of a conchal cartilage (ipsilateral, contralateral, or both) or a costal cartilage framework depends on the size and extent of the cartilage defect (see Fig. 33-1, *A* through *C*). This choice is usually clear before the procedure begins; a template of the normal ear is used as a guide to assess the extent of the defect. Dissection of the overlying skin is best (and most safely) accomplished with an incision just along the edge of the helical rim, with the anterior skin dissected as far as the conchal border (fully exposing the limits of the contracted scapha and anti-helix), and the postauricular skin is dissected to the extent that the mastoid fascia can be accessed for placing supporting sutures⁶ (Fig. 33-2). At this point, the dissection continues to expose the

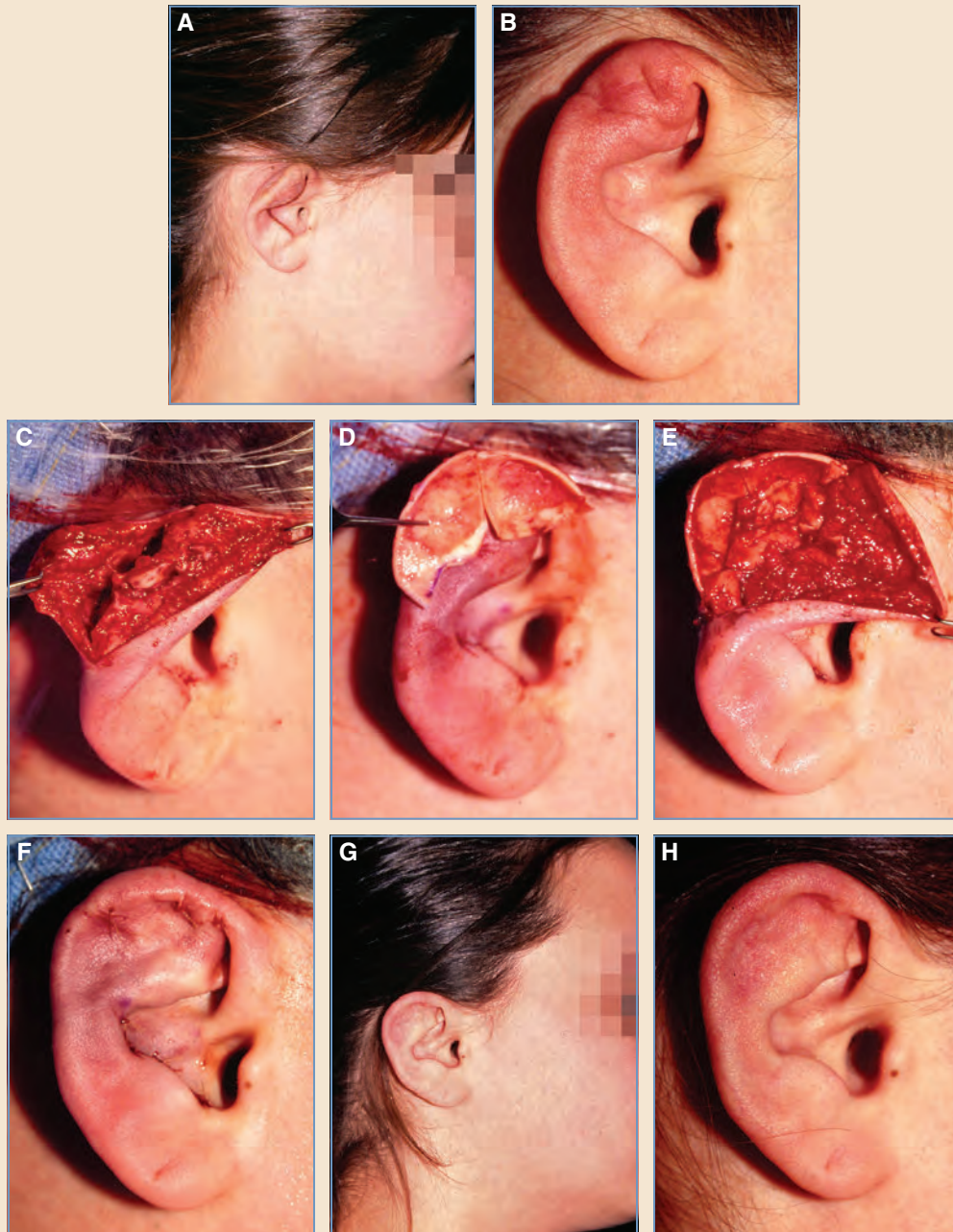


Fig. 33-2 Reconstruction of a postchondritis deformity. **A** and **B**, Nine months after resolution of an infection that occurred after ear piercing. **C**, Meticulous dissection of the skin flap is required to fully view the deformity and cartilage loss from the helical rim and scapha. **D**, Ipsilateral and contralateral conchal cartilage grafts are positioned where they will be spliced into the remnant. **E**, The skin flap is redraped and fixed to the cartilage with a variable number of fine through-and-through chromic sutures. **F**, Six months after reconstruction. **G** and **H**, Full lateral and closeup views 6 months after reconstruction.

remaining borders of the normal helical crus and helical rim at the borders of the deformed cartilage and normal landmarks. The grafts are then placed, fixing them to the remaining portions of the antihelical body, helical crus at the helical root, and midhelical rim with 5-0 and 6-0 clear nylon. Supporting sutures are placed from the area of the reconstructed scapha and triangular fossa to the temporal and mastoid fascia as the graft with 4-0 clear nylon; these sutures should be placed when the grafts are placed to more easily position the grafts and shape the new upper pole. Although a conchal graft of virtually the whole conchal floor can be harvested as a graft from the contralateral ear, the surgeon should limit the size of ipsilateral conchal graft to ensure that the structural support of the body of the antihelix is not lost. This will still allow a reasonable-sized graft to augment the graft harvested from the contralateral side.

Traumatic Deformities With Limited Skin Loss or Scarring and Segmental Cartilage Loss

The next subgroup of acquired deformities, which still falls within the group of skin and cartilage deficiency with limited scarring, involves traumatic deformities in which upper, middle, or lower pole tissue loss has been limited to the ear without scarring of the postauricular and mastoid skin. Here there is a wide enough area of non-hair-bearing postauricular skin available to cover the replaced missing segment of cartilage (see Fig. 33-2, *D* and *E*). In this group the postauricular and mastoid skin can be elevated in continuity with the border of the defect (as a broad bipediced flap); the cartilage framework is placed beneath this skin and spliced to the remaining auricular remnant, and the ear is elevated in a second stage, with a skin graft applied to the postauricular surface and sulcus (Figs. 33-3 and 33-4).

Incomplete or Failed Reconstruction

Another subgroup of patients that may fit within this broad classification of acquired deformities includes those who have had incomplete or failed reconstruction of microtia or constricted ear deformities.^{7,8} Roughly speaking, if one is seeing more than the rare ear reconstruction patient seeking reoperation, this subset of patients can be further divided into (1) those who have had what can best be described as a rearrangement of the microtic vestige in early childhood but little else until their presentation years later, (2) those who have been through a standard staged reconstruction with a costal cartilage framework who have lost part of the framework because of infection or partial flap compromise with no additional efforts to correct the deformed auricle, and (3) those in whom the initial reconstruction failed and a fascial flap salvage was attempted, which subsequently also failed.⁷⁻⁹ These patients have all of the varied tissue requirements seen in the decision-making process with other acquired deformities.



Fig. 33-3 A, This boy lost the middle third of his left ear at age 14, including the helical rim and a segment of the antihelix, from a dog bite. B and C, The ear 1 year after the injury and just before surgery. D and E, Intraoperatively the defect and the pattern showing the missing segment. F, The result 1 week after coverage of the combined ipsilateral and contralateral conchal grafts with a flap elevated on the border of the defect. G-I, The result 1 year after surgery shows that although the midhelical rim is not prominent on either ear, the symmetry is excellent, particularly as seen in the posterior view.

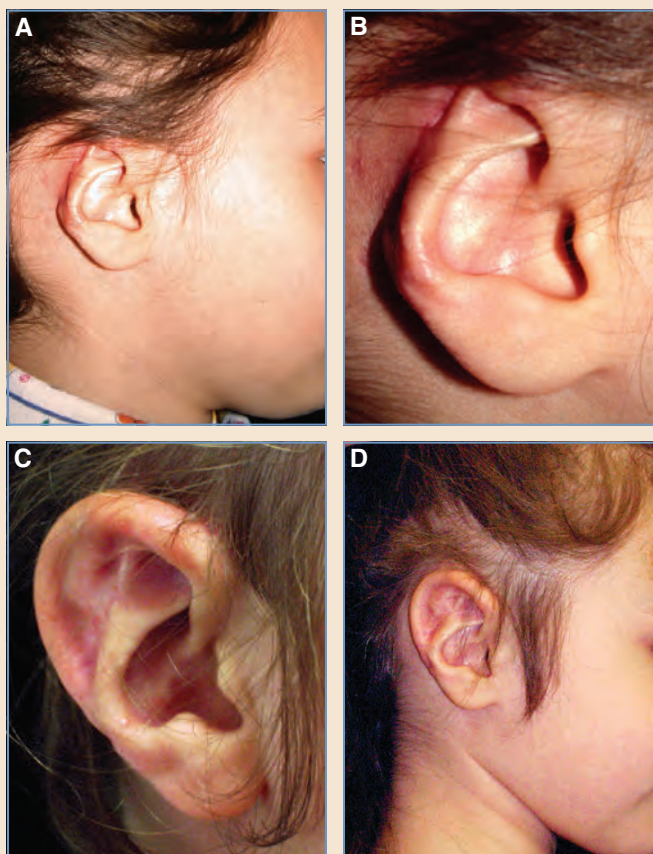


Fig. 33-4 This 5-year-old girl lost the upper half of her ear when she was bitten by a dog. **A** and **B**, There was no significant scarring on the adjacent postauricular skin, which still included a relatively wide zone of non-hair-bearing skin. This skin was elevated in continuity with the border of the defect as a bipedicle flap to allow splicing of a costal cartilage framework to the existing auricular cartilage. **C** and **D**, Reconstruction was completed with elevation of the ear and reconstruction of the sulcus with a full-thickness skin graft. Results are shown 1 year postoperatively.

It seems intuitive that patients in the first group are good candidates for placing a costal cartilage framework through an approach similar to the typical reconstruction of a lobular or conchal remnant type of microtia with minor variations (Fig. 33-5). However, it may be less obvious that some patients in the second group may also be candidates for a new cartilage framework placed into the same skin envelope. For this group of patients, I (B.S.B.) might previously have used a temporoparietal fascial flap (TPFF) and skin graft for total replacement of the skin envelope; however, following examples demonstrated by Firmin and Marchac,⁵ I (B.S.B.) now recognize that this is not only unnecessary, but also it is often a more extensive procedure than required. If these cases are well selected, the skin coverage is more delicate and has a better color match than a skin-grafted TPFF, and the additional donor scar in the scalp is avoided.

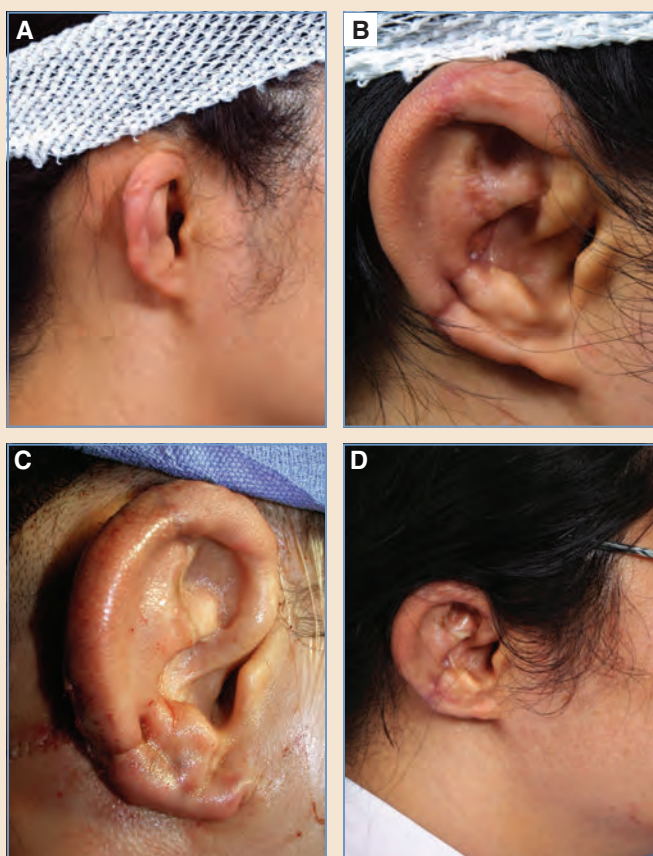


Fig. 33-5 A, This 34-year-old man, who had undergone a single unsatisfactory attempt to reconstruct a conchal remnant type of microtia in childhood, underwent reconstruction through an approach similar to a standard reconstruction. B, After first-stage reconstruction, he still had a relatively limited amount of lobular tissue. C, The ear was elevated and the lobule augmented with a dermal fat graft. D, The result 2 years after further augmentation of the lobule and minor additional soft tissue revision.

What distinguishes skin-grafted TPFF is the thin, supple quality of the skin, which moves freely over the surface of the amorphous framework below. In patients with a skin-grafted TPFF, the skin can be meticulously elevated off the framework through an incision along the helical rim (typically at the junction with the postauricular skin graft), the unacceptable framework removed, the tethering scars released, and the new framework placed (Fig. 33-6). The postauricular graft can typically be raised with some underlying fascia as a flap to add to the overall envelope and in some patients to allow some depth to the postauricular sulcus from the outset. There is a limit to how much conchal excavation can initially be accomplished without compromising the vascularity of the skin envelope. The primary goal of the first reoperation is to place a well-sculpted framework beneath the thin, delicate, flexible skin envelope and to correct any malposition of the earlier reconstruction. After the new framework is in place, subsequent procedures can excavate the concha, construct the tragus, and further define the postauricular sulcus according to the techniques used in a typical successful staged reconstruction.

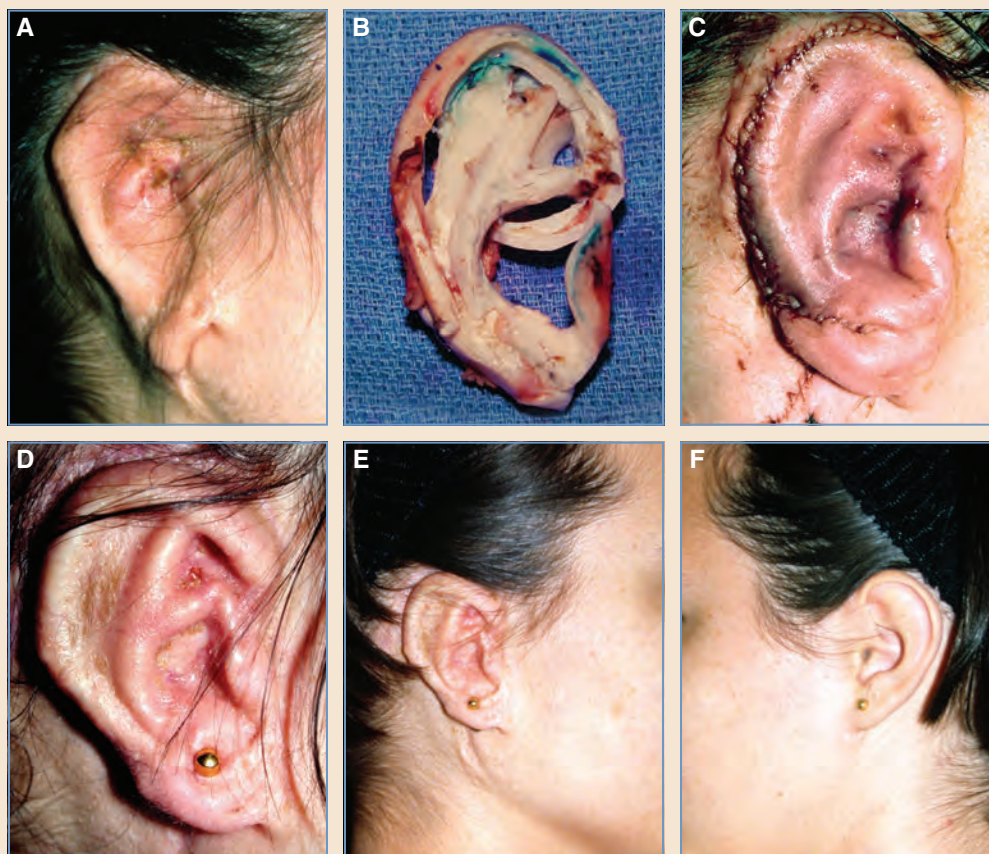


Fig. 33-6 This 12-year-old girl had undergone multiple unsuccessful reconstructive procedures for a lobular type of microtia. **A**, Despite multiple surgeries and scarring, the skin was still thin and supple and could be used to cover a new cartilage framework. **B** and **C**, The new framework was constructed and placed through an incision along the helical rim, which allowed resection of the old framework and maintained the vascular supply to the flap. **D**, Closeup after elevation of the ear was completed. **E** and **F**, Completed repeat reconstruction and contralateral ear 2 years postoperatively.

Tissue Expansion in the Treatment of Acquired Defects With Limited Scarring

One additional approach that should be discussed briefly before leaving the group of skin and cartilage deformities with limited scarring is the role of tissue expansion in providing additional skin. Some patients present with skin that is deficient but not severely scarred and in whom previous reconstructive efforts have not disrupted the adjacent skin with multiple scars and skin grafts. Several investigators have successfully used tissue expansion in this group of patients^{10,11}; however, the potential risks of expansion (particularly expander exposure) and the need to at least partially excise the expander capsule to ensure adequate skin/cartilage coaptation and vascular ingrowth to the graft may increase the likelihood of flap compromise. In addition, it is rarely possible to expand the skin sufficiently to avoid additional grafting to the postauricular sulcus, which is a benefit frequently mentioned as the primary indication for tissue expansion in these reconstructions. One final caveat is that if one chooses to use tissue expansion for reconstruction of the ear, critical tissues must never be compromised in an effort to salvage an exposed expander. If this

occurs, the expander should be removed, the tissues allowed to settle, and an alternative approach selected to complete the reconstruction.

From a timing standpoint, the tissues and scars need a chance to settle and mature before the reconstruction is initiated. This is particularly important when the skin flap used in the reconstruction will be based along the margin of the defect. In our opinion, the surgeon should wait at least 6 to 12 months from the time of injury or resection, although the amount of time may vary on a case-by-case basis. For patients whose cartilage loss resulted from chondritis, the surgeon should wait until the skin feels soft and mobile. For patients with skin loss and surrounding soft tissue injury, a waiting period of at least 1 year is often prudent. However, if the plan is for reconstruction with a TPFF, the periauricular tissues are uninjured, and the remaining ear is relatively uninjured, an earlier reconstruction can be considered.

SKIN AND CARTILAGE DEFICIENCY WITH MODERATE AND SEVERE SCARRING

Patients who have skin and cartilage deficiency with moderate and severe scarring typically present with either a history of major trauma to the ear and surrounding face and scalp or a history of multiple surgeries with episodes of infection, flap loss, cartilage exposure, and multiple regional flaps for attempted salvage. Often these regional flaps, even if they are performed successfully, may have been delayed long enough, before attempted salvage of the previously failed flap, for the vascularity of the cartilage framework to be compromised; subsequently the outcome is complete cartilage resorption, or significant enough resorption to lose framework integrity. In these patients it is necessary to provide vascular coverage that has not been previously compromised by the previous injury, and coverage with vascularized fascial flaps is the treatment of choice.

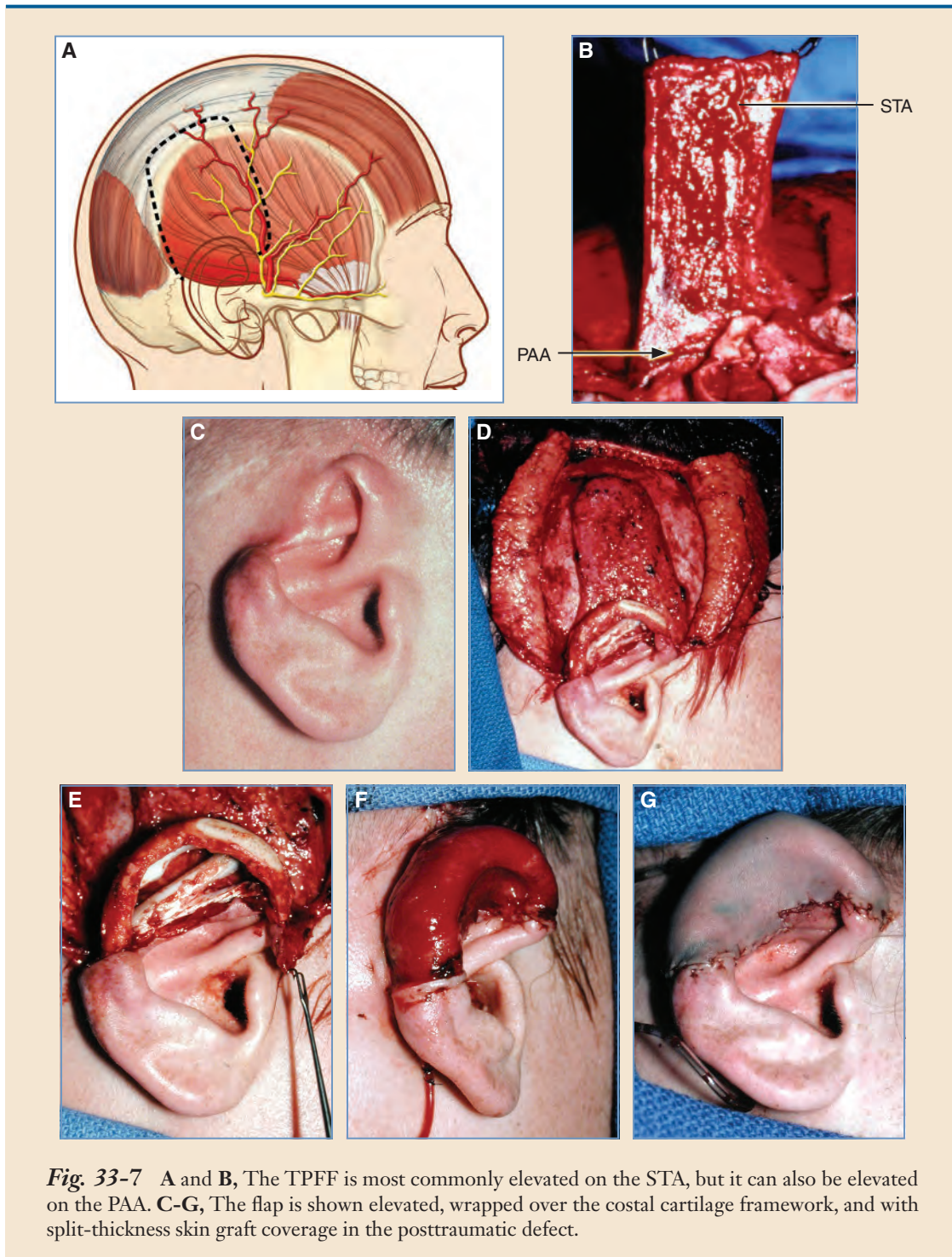
Before we discuss specific cases, it is appropriate to specifically look at the role of fascial flaps in ear reconstruction.

Fascial Flaps in Ear Reconstruction

Fascial flaps, although more typically used in treating major acquired deformities of the ear, have had a role in the treatment of microtia cases associated with auricular dystopia and more significant degrees of craniofacial microsomia; they are also used in salvaging complications of microtia reconstruction where scarring obviates the use of the regional skin (Figs. 33-7 through 33-9). A small fascial flap may provide a single-stage reconstruction of a small segmental defect, but it is most often held as a fallback for reconstruction of more complex defects.

Temporoparietal Fascial Flap

The TPFF is the most commonly used of the fascial flaps in complex ear reconstruction, although the postauricular fascial flap and deep temporal fascial flap may occasionally be used. The TPFF may also be used as a free flap when previous trauma or iatrogenic injury has compromised the ipsilateral fascial vascular supply. The flap is supplied by both the superficial temporal artery (STA) and postauricular artery (PAA), and although the flap is typically elevated on the STA alone, both vessels should be included to minimize the risk of partial flap necrosis whenever possible. Although the TPFF has also been used as a random flap by Brent and Byrd,¹² this type of flap is generally not recommended.



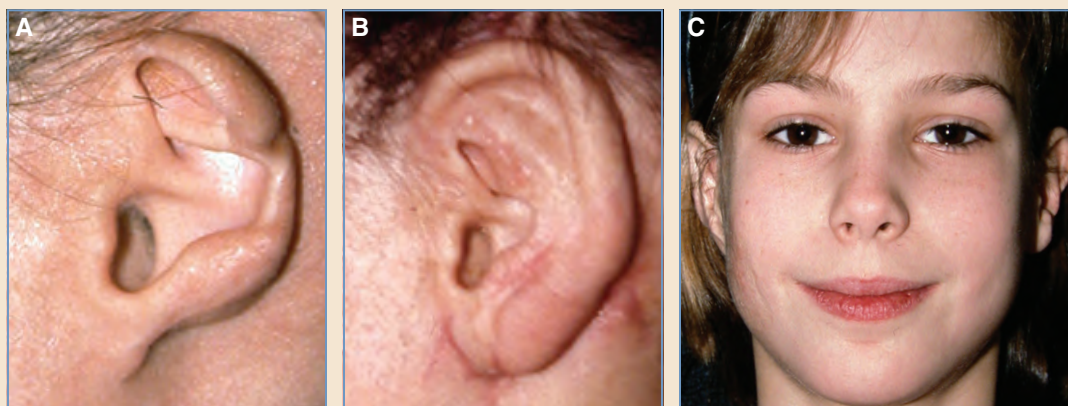


Fig. 33-8 A, Extensive loss of the greater part of the ear, with extensive scarring surrounding the ear and extending onto the cheek, appeared to be the result of this young girl's intrauterine vascular accident, which resulted in compromise to the ear blood supply and loss of both ear skin and cartilage. B, Reconstruction was carried out with a costal cartilage framework covered with a TPF and a split-thickness skin graft. She is shown 1 year after a two-stage reconstruction. C, One year postoperatively.

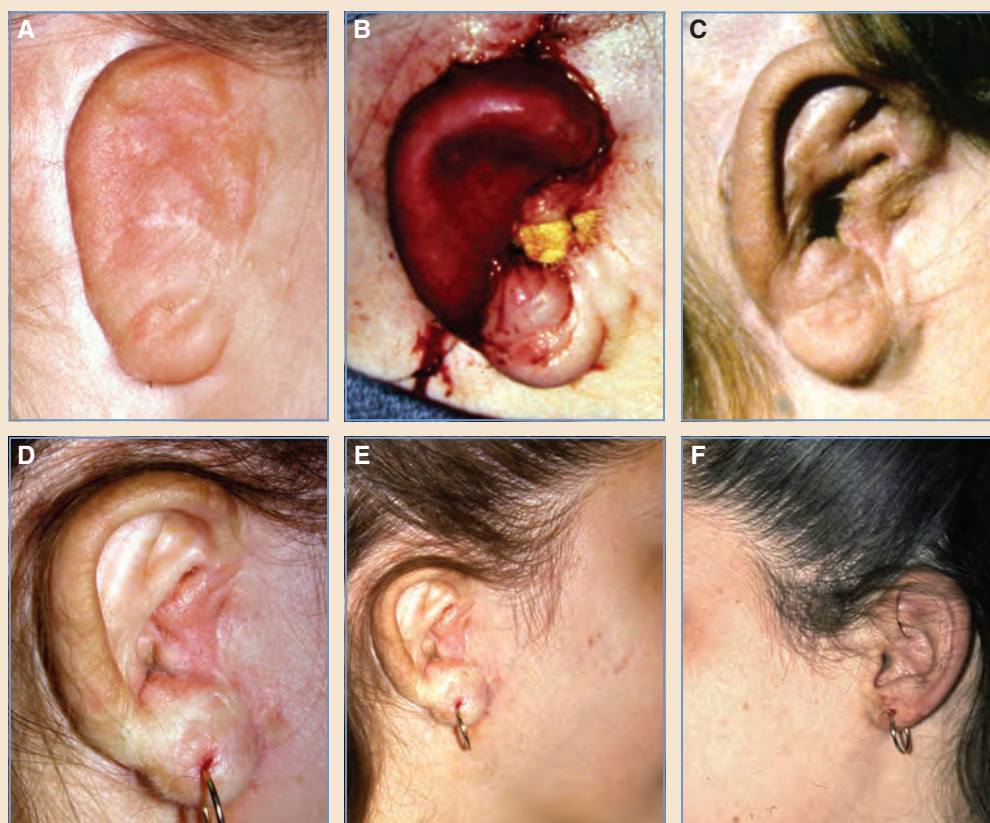


Fig. 33-9 A, This 7-year-old girl had undergone seven previous attempts at reconstruction of a lobular type of microtia, which resulted in a small amorphous ear. B, A new framework was placed and covered with a TPF and split-thickness skin graft, retaining enough of the original tissue to create the tragal prominence and lobule. C, At 3 months postoperatively (after the first stage of repeat reconstruction), the fascial flap was thinning and the details of the ear were more evident. D, Result of the one-stage repeat reconstruction, seen 6 years later. E and F, Symmetry with the contralateral normal ear can be seen.

The postauricular fascial flap supplied by the PAA and occipital arteries is a continuation of the TPFF (superficial temporal fascia). The deep temporal fascial flap is supplied by the middle temporal artery, which arises from the STA.^{8,9} It may also include a branch of the PAA. Both of these flaps may play a role in secondary or reoperative ear surgery, particularly as salvage after failure of the TPFF. Although the former is quite effective for coverage of small areas of skin or flap loss in the middle and lower third of the ear (and on occasion is even preferred over the TPFF), these other flaps are used relatively rarely.

If a previous failure to replant and attempt salvage has not compromised the STA, or at least has left a clearly defined length of the PAA and occipital arteries, and there is still donor costal cartilage available, the surgeon can consider discarding most or all of the previous ear vestige. An effort should be made to keep the usable parts of any ear remnant as would be done when reconstructing a conchal remnant microtia. At the very least the surgeon should retain enough tissue to reconstruct the lobule^{3,13,14} (see Fig. 33-9).

Careful assessment of the surrounding scars (including careful inspection for any hidden scalp scars) and mapping potential vascular pedicles with a Doppler probe are key factors in performing these complex reconstructions. I (B.S.B.) have seen more than a few patients in whom there was a failed reconstruction or patients who had undergone prior procedures in the early postinjury period that left scars within the scalp at surgical sites that defied logic and that further complicated reconstruction. The TPFF is the most commonly used fascial flap for complex ear reconstruction, and in some cases the surgeon may need to be innovative in its design while still respecting the vascular anatomy^{3,13,14} (Fig. 33-10). Alternative choices include the postauricular fascial flap and free tissue transfer of the TPFF from the contralateral side⁷⁻⁹ (Fig. 33-11).

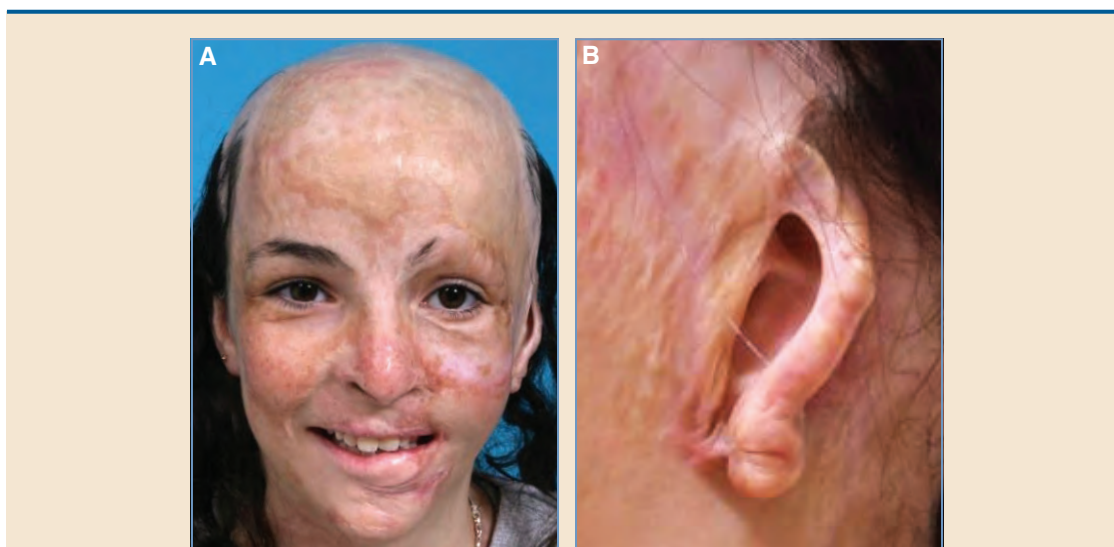


Fig. 33-10 A and B, At age 10 years this girl sustained extensive burns in a terrorist attack, resulting in the loss of most of her scalp and the greater part of her left ear.

Continued

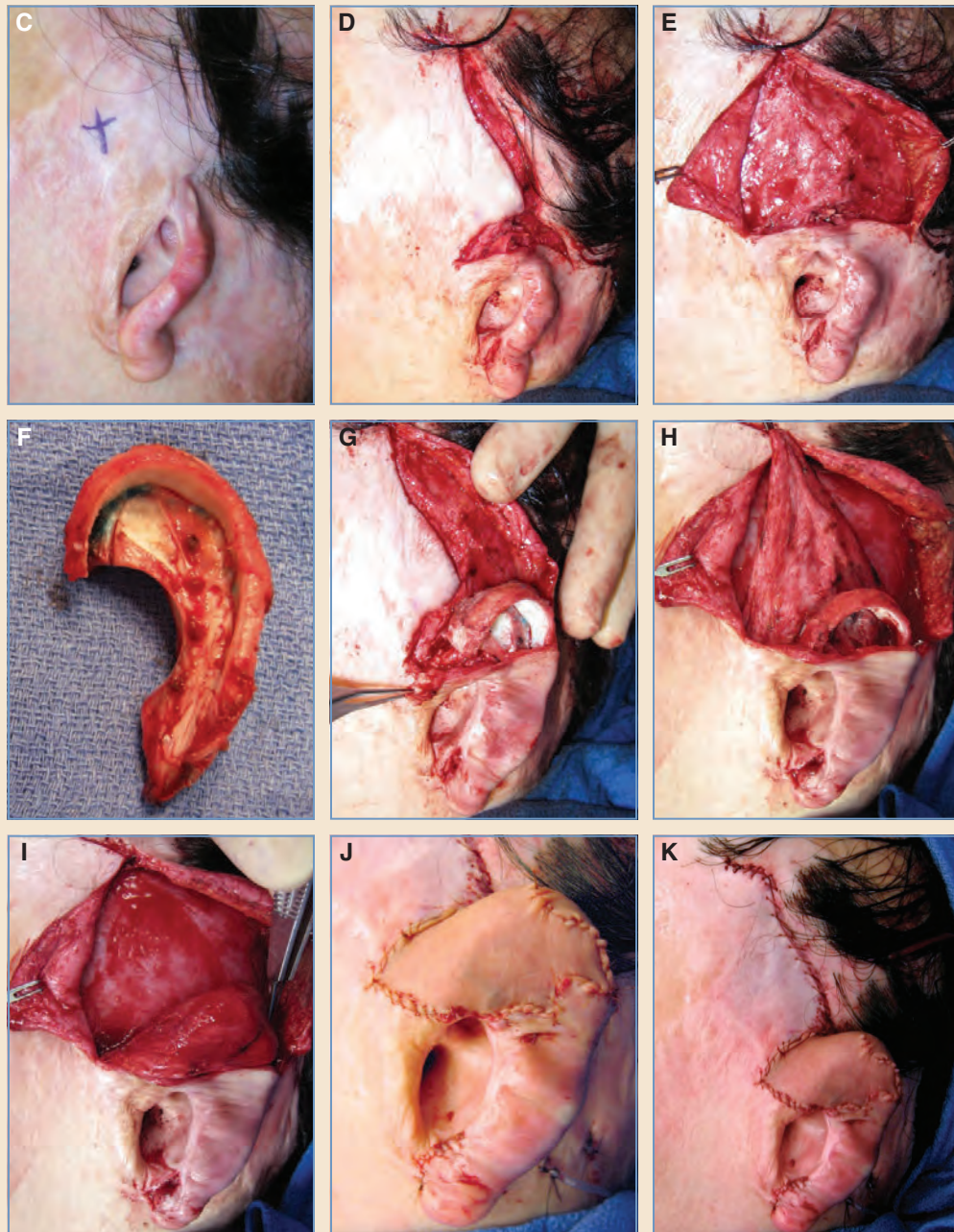


Fig. 33-10, cont'd C-K, To avoid potential loss of the limited remaining normal scalp, the incisions were oriented to preserve the scalp and allow adequate skin flap elevation off the remaining cartilage vestige and thin coverage of the framework as it was spliced into place. The TPF was oriented on the anterior branch of the STA and covered the upper pole of the reconstruction. The greater part of the reconstructed framework was covered by the thin and pliable postauricular skin that was elevated in continuity with the border of the defect. The fascial flap was then covered with a split-thickness skin graft.



Fig. 33-10, cont'd **L**, The result 4½ years after first-stage reconstruction. **M**, The result 2 years after second-stage reconstruction, which involved scar revision and the insertion of dermal fat grafts into the lobule. At the second-stage reconstruction, she also underwent brow reconstruction with micro hair grafts. Her insufficient normal hair-bearing scalp prevents scalp reconstruction with either tissue expansion or micrografting.



Fig. 33-11 **A-E**, This 18-year-old girl sustained extensive soft tissue injuries, including the loss of the soft tissue of her lateral cheek, the greater part of her left ear, and the underlying superficial temporal fascia. She underwent a series of reconstructive surgeries, including free tissue transfer, which provided a lobular segment of tissue usable for earlobe reconstruction but was positioned anterior and inferior to the planned reconstruction.

Continued





Fig. 33-11, cont'd M and N, Seven months after first-stage reconstruction, the second stage involved lobule and tragus construction from anteriorly displaced remnants and banked “tragal” cartilage grafts. O and P, Two weeks after second-stage reconstruction. Q, Four years after first-stage reconstruction and before the final stage including facial scar revision. R and S, Three months after final-stage ear reconstruction.

Grafted Fascial Flap

Staging the reconstruction is similar to that described for complex microtia cases in which a fascial flap is required for vascular coverage. Templates are made for the framework and placement of the ear. Cartilage is harvested and the framework is constructed. The framework is placed and covered with the fascial flap, and the flap is covered with a split-thickness skin graft, with the scalp the preferable donor site for the graft. Vacuum drains are placed beneath the framework and flap and beneath the donor scalp flaps. The postoperative treatment is similar to that used for standard reconstruction, but the time between the first (new reconstructive stage) and subsequent stages may be longer than with skin flap coverage. Grafted fascial flaps are quite variable in the time course of thinning to show the underlying cartilage details. Regardless of the progress, I (B.S.B.) often wait at least 6 months between the first and subsequent stages. Patients must be aware that in some cases, the thinning may take as long as 1 year before the framework details are well defined. One can expect continued thinning along with improvement of skin color and flexibility of this flap over at least 2 to 3 years, if not longer. If the surgeon has properly prepared these individuals for their repeat reconstruction, they generally are accepting of the slow healing process and are patient enough to look toward the final result, even if it takes months or years (see Figs. 33-9 through 33-11).

PROSTHETIC RECONSTRUCTION

Not surprisingly, the success of reconstruction of acquired deformities, regardless of cause, depends significantly on careful case selection. Most often, except for individuals with previous limited surgery and limited scarring, the surgeon should consider and discuss the possibility of prosthetic reconstruction. At the very least, any patient who has a history of multiple previous ear reconstructions and is dissatisfied with the size, shape, position, and lack of fine detail in the outcome should be evaluated for prosthetic reconstruction.^{15,16} This includes patients who have had previous reoperative surgery with cartilage harvested from both sides of the chest and patients who have lost the ear as a result of tumor resection who require long-term monitoring for possible tumor recurrence. Patients should meet with both the surgeon involved in the osteointegration of the abutment for the prosthetic ear (if it is not you) and the anaplastologist who will be creating the prosthesis. They should understand not only the obvious benefits of an excellent match in size, shape, and color with the current prosthesis, but also the long-term need to replace the prosthesis, maintenance of the abutment, and the costs of prosthetic replacement.

In my practice, even in patients in whom I (B.S.B.) am confident of the success of an autologous reconstruction, I (B.S.B.) evaluate the patient for a prosthetic reconstruction and discuss this option, because this ensures that he or she is making an informed decision when proceeding with a new set of staged procedures. This routine also provides a means of informing the patient of an additional “final option” if reconstruction is unsuccessful (Fig. 33-12).



Fig. 33-12 A, This young boy had an unusual sarcoma involving his right ear. He had had multiple recurrences and ultimately the ear was removed. His mother was a nurse and did not want an autologous reconstruction, because she wanted to watch for recurrences. B, The prosthesis fabricated for the reconstruction. C, The osteointegrated prosthesis in place, which he has worn for 15 years. (Courtesy of Gordon H. Wilkes, MD.)

CONCLUSION

Successful staged reconstruction of the ear for major acquired ear deformities requires meticulous attention to all aspects of the available auricular and donor tissue and the soft tissue environment surrounding the planned reconstruction. Because there is considerable variation in auricular remnants, meticulous planning is perhaps the most important factor in reducing complications.

The challenges faced during successful reconstruction of “virgin microtia,” or the first attempt at reconstruction of an acquired ear deformity, are significantly magnified when these procedures have already failed and the patient presents for reoperation. Secondary surgery to correct a previously unsatisfactory result must take into account that the recipient bed is now scarred and the

donor tissues are more limited. Successful reoperation requires familiarity with all available reconstructive options and the expertise of the operating surgeon. The surgeon must ensure that the patient understands the complexity of the surgery and that the reconstruction will probably be staged. The surgeon must also be reasonably certain that the patient's expectations and the surgeon's capabilities are in sync. Finally, the alternatives of accepting the current result at one end of the spectrum and prosthetic reconstruction at the other end must always be considered and discussed. With these issues in mind, the surgeon, with consummate skill and a little good luck on occasion, will see that the patient does not require additional reoperation.

KEY POINTS

- The principles of treating congenital ear deformities are directly translatable to the treatment of acquired ear deformities.
- Classifying acquired ear deformities into two groups—those with skin and cartilage deficiency with limited scarring and those with skin and cartilage deficiency with moderate-to-severe scarring—allows a surgeon to develop a readily applied treatment algorithm for any and all acquired deformities.
- Careful appraisal of the defect, donor tissue, and the surrounding soft tissue is imperative to selecting the optimal reconstructive modality.
- When correcting an acquired ear deformity, a critical decision must be made: to reconstruct to match the size of the opposite ear or to normalize the shape of the reconstructed ear and accept a size difference.
- Because both ears are rarely seen at the same time, differences of 0.5 to 1.0 cm may not be very noticeable, and simpler procedures may often produce a better shape without matching vertical height.
- Fascial flaps are important sources of tissue that can be used in salvage or secondary reconstruction procedures, or when local skin flaps cannot achieve adequate coverage of the auricular framework.
- All treatment options, including prosthetic reconstruction or potentially accepting the ear in its defected state, should be considered and thoroughly discussed before initiating treatment.

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34

Congenital and Acquired Deformities of the Eyelids

Milap Mehta • Bruce S. Bauer



eyelids exist to protect the eye, the most complex of human sensory organs. Their multilamellar structure provides glands that lubricate and irrigate the cornea, on whose transparency we rely for our detailed view of the world around us. The fibrocartilaginous tarsal plates provide delicate structural support, and the opposing orbicularis oculi and levator palpebrae muscle systems allow rapid blink and reflex closure to protect against particles or insects headed toward the cornea.

The development of visual acuity in newborns depends on the reception of visual images on an infant's retina. If growths or abnormalities in or around the eyelids prevent reception of these images, vision will not develop in the eye, a condition known as *amblyopia ex anopsia*. For this reason, a condition such as cryptophthalmia (fusion of the eyelids over an otherwise normal eye) or a hemangioma of the eyelid that completely blocks vision should be treated as a medical emergency. Similarly, an infant with a coloboma of the upper eyelid, or extreme proptosis, should be treated immediately to prevent corneal scarring or perforation and loss of the eye. After the period of visual development, the protection of the globe becomes the paramount function of the eyelids. Trauma with laceration or avulsions of the eyelid, often associated with facial fractures, will require careful and complete repair to preserve the eye.

DEVELOPMENT

The clinical significance of understanding these developmental processes cannot be underestimated, because it may help to understand the mechanisms of the development of many congenital anomalies. The development of the eye begins with a thickening of the diencephalic neural folds at 22 days' gestation. The eyelids form as ectodermal folds between week 6 and week 8 of development. As the lid folds advance and fuse, uniting by week 12 to week 14, they form an internal

pocket that covers the eye and differentiates on the deep surface to form the conjunctiva and its adnexa, including, at 6 to 8 weeks of age, the lacrimal gland.^{1,2}

The lacrimal drainage system develops as an ectodermal cord between the lateral nasal process and the maxillary process. The epithelial cord canalizes from month 3 to month 6 of gestation. The punctum remains closed until the month 7, and the nasolacrimal duct may not canalize until just before birth. Canalization of the nasolacrimal duct may be incomplete in up to 30% of newborns, but only a small number of these become symptomatic, because the duct usually opens spontaneously.

Subsequent development of the tarsal plates, lash follicles, and meibomian, Zeis, and Moll glands occurs until about weeks 22 through 24 of gestation. At that time, or within weeks after, the fused lids separate into the upper and lower eyelid.

Alterations in these developmental processes can lead to a wide variation of anomalies,^{3,4} as shown in Table 34-1. For example, colobomas and clefts may occur from inadequate fusion of the

Table 34-1 Some Syndromes With Ocular Abnormalities

Syndrome	Comments (chromosome map locations)
Aicardi syndrome	Lacunar retinopathy Probably X-linked dominant; lethal in male hemizygotes (Xp22)
Alagille syndrome	Posterior embryotoxon, pigmentary retinopathy Autosomal dominant; <i>JAG1</i> mutations (20p12)
Anophthalmia-syndactyly syndrome	Autosomal recessive
Apert syndrome	Ocular proptosis
Baraitser-Winter syndrome	Iris coloboma, ptosis, hypertelorism Autosomal recessive
Biemond syndrome II	Iris coloboma Autosomal dominant
Blepharophimosis-ptosis-epicanthus inversus	Autosomal dominant; <i>FOXL2</i> mutations (3q23)
CHARGE association and Hall-Hittner syndrome	Colobomas, sporadic, some familial cases
CHIME syndrome	Colobomas Autosomal recessive
Chondrodysplasia punctata	Cataracts Genetically heterogeneous Conradi-Hünermann type, X-linked dominant; <i>EPB</i> mutations (Xp11.22-p11.23) Rhizomelic type, autosomal recessive; <i>PEX7</i> mutations (6q22-q24)
Cohen syndrome	Retinal mottling, constricted visual fields, other ocular abnormalities Autosomal recessive; <i>COH1</i> mutations (8q22-q23)
Crouzon syndrome	Ocular proptosis
Cryptophthalmos syndrome	Autosomal recessive; <i>FRAS1</i> mutations (4q21)
Delleman syndrome	Orbital cysts, microphthalmia, colobomas All cases sporadic, to date

CHARGE, Coloboma, heart defect, atresia choanae, retarded growth and development, genital abnormality, and ear abnormality. *CHIME*, coloboma, heart defects, ichthyosiform dermatosis, mental retardation, and ear anomalies with hearing loss.

Syndrome	Comments (chromosome map locations)
Ehlers-Danlos syndrome, type VI	Keratoglobus, other ocular abnormalities Autosomal recessive
Fabry syndrome	Aneurysmal dilation and tortuosity of conjunctival and retinal vessels, characteristic corneal and lenticular alternations X-linked recessive; <i>GLA</i> mutations (Xq22)
Familial dysautonomia	Decreased tears, absent corneal reflex, other ocular abnormalities Autosomal recessive; <i>IKBKAP</i> mutations (9q31)
Fetal alcohol syndrome	Narrow palpebral fissures, small eye globes
Focal dermal hypoplasia	Colobomas X-linked dominant, lethal in male hemizygotes
Goldenhar syndrome	Epibulbar dermoids, other ocular abnormalities Most cases are sporadic, some have autosomal dominant inheritance
Hallermann-Streiff syndrome	Congenital cataracts, microphthalmia, other ocular abnormalities All cases are sporadic
Holoprosencephaly	Ocular hypotelorism, single eye globe in cyclopia, other ocular abnormalities Causatively heterogeneous
Homocystinuria	Ectopia lentis (inferior dislocation), other ocular abnormalities Autosomal recessive; <i>CBS</i> mutations (21q22.3)
Kaufman syndrome	Upslanting palpebral fissures, sparse eyebrows, blepharophimosis, myopia, microcornea, pale optic disks
Lenz microphthalmia syndrome	Microphthalmia, anophthalmia X-linked (Xq27-q28)
Marden-Walker syndrome	Blepharophimosis, other ocular abnormalities Autosomal recessive
Marfan syndrome	Ectopia lentis (superior dislocation), other ocular abnormalities Autosomal dominant; <i>FBNI</i> mutations (15q21.1)
Marshall syndrome	Myopia, cataracts Dominant inheritance, autosomal dominant versus X-linked dominant Basic defect is controversial; <i>COL1A1</i> (1p21) may be a candidate gene, but this has been debated.
Megalocornea and mental retardation	Four types: Neuhäuser syndrome, Frank-ter Haar syndrome, Verloes syndrome, and Frydman syndrome
Michels syndrome	Blepharophimosis, ptosis, epicanthus Autosomal recessive
Moebius syndrome	Seventh nerve palsy If syndrome is defined as involvement of both sixth and seventh nerve palsies, almost all cases are sporadic.
Mucopolysaccharidoses	Corneal opacities in types I-H (Hurler), I-H/S (Hurler-Scheie), I-S (Scheie), IV-A (Morquio A), IV-B (Morquio B), VI-A (Maroteaux-Lamy A), and VI-B (Maroteaux-Lamy B)
Nance-Horan syndrome	Congenital posterior cataracts X-linked (Xp22.13-p22.31)
Neurofibromatosis	Type I Lisch nodules, occasionally pulsating exophthalmos caused by bony defect of posterosuperior orbital wall, uncommonly optic glioma Caused by <i>NF1</i> mutations (17q11.2) Type II Senile lens opacities or subcapsular cataracts Caused by <i>NF2</i> mutations (22q12.2) Types I and II: autosomal dominant

Continued

Table 34-1 Some Syndromes With Ocular Abnormalities—cont'd

Syndrome	Comments (chromosome map locations)
Osteogenesis imperfecta	Blue sclera in types II and III <i>COL1A1</i> mutations (17q21.31-q22) or <i>COL1A2</i> mutations (7q22.1) in both types II and III
Peters-plus syndrome	Consists of central corneal opacity, thinning of the posterior cornea, and iridocorneal adhesions Also found: anterior chamber defects and other ocular anomalies Autosomal recessive; a mutation in <i>PAX6</i> (11p13) in one case and a mutation in <i>RIEGL</i> in another
Pfeiffer syndrome	Ocular proptosis
Rieger syndrome	Abnormal development of the anterior chamber of the eye Autosomal dominant; <i>RIEGL</i> mutations (14q25-q26)
Rubella syndrome	Cataracts
Stickler syndrome	Type I High myopia with retinal detachment, cataracts Autosomal dominant; <i>COL2A1</i> mutations (12q13.11-q13.2) Type II Severe, early onset and retinal degeneration Autosomal dominant; <i>COL1A1</i> mutations (1p21)
Trisomy 21 syndrome	Upslanting palpebral fissures, epicanthic folds, Brushfield spots, fine lens opacities, convergent strabismus, nystagmus, keratoconus, cataract, rare retinoblastoma
Trisomy 13 syndrome	Microphthalmia, iris coloboma, retinal dysplasia
Trisomy 18 syndrome	Various abnormalities of the eye globe, adnexa, and neuroophthalmologic system
Tuberous sclerosis	Retinal hamartomas Autosomal dominant; mutations in either <i>TSC1</i> (9q34) or <i>TSC2</i> (16p13.3)
Velocardiofacial syndrome	Narrow palpebral fissures, blue suborbital coloring, tortuous retinal vessels, posterior embryotoxon, small optic disks, cataracts, colobomas Autosomal dominant (22q11.2)
Waardenburg syndrome	Type I Heterochromia irides, dystopia canthorum Autosomal dominant; <i>PAX3</i> mutations (2q35) Types IIA and IIB Heterochromia irides without dystopia canthorum Type IIA: autosomal dominant; <i>MITF</i> mutations (3p12.3-p14.1) Type IIB: autosomal dominant (1p13.3-p21) Type III Klein-Waardenburg syndrome, heterochromia irides, dystopia canthorum, limb defects Autosomal dominant; <i>PAX3</i> mutations (2q35) Type IV Waardenburg-Shah syndrome, heterochromia irides without dystopia canthorum, associated Hirschsprung disease Causatively heterogeneous Mutations in <i>EDHRB</i> (13q22) or in <i>EDN3</i> (20q13.2-q13.3); both are autosomal recessive; or in <i>SOX10</i> (22q13), autosomal dominant
Wagner syndrome	Hyaloideretinal degeneration, chorioretinal atrophy, cataract, peripheral retinal detachment, glaucoma Autosomal dominant (5q13-q14)
WAGR	Wilms tumor, aniridia, gonadoblastoma, mental retardation del(11)(p13)
Walker-Warburg syndrome	Microphthalmia and many other ocular abnormalities Autosomal recessive; <i>POMT1</i> mutations (9q34.1) and <i>FCMD</i> mutations (9q31)

Syndrome	Comments (chromosome map locations)
Weill-Marchesani syndrome	Spherophakia Causatively heterogeneous Autosomal dominant form with <i>FBNI</i> mutations (15q21.1) and autosomal recessive form (19p13.2-p13.3)
Werner syndrome	Senile cataracts Autosomal recessive; <i>RECQL2</i> (8p11.2-p12)
Williams syndrome	Stellate iris, strabismus Contiguous gene syndrome involving <i>ELN</i> (7q11.2)
Wilson disease	Kayser-Fleischer rings Autosomal recessive; <i>ATP7B</i> mutations (13q14.3-q21.1)

Table 34-2 Duke-Elder's Pathology of the Eyelids

Stage of Development	Pathology	Conditions
First trimester (early)	Failure of formation	Cryptophthalmos Microphthalmos Colobomas
Second trimester (5 to 6 months)	Failure of formation	Ankyloblepharon Congenital ectropion Entropion
Third trimester (late)	Failure of differentiation	In the tarsus, palpebral musculature, and lid skin

processes of the embryonic lid folds. Cryptophthalmos is caused by failure of the lid folds to develop, whereas anophthalmos or microphthalmos may result from abnormalities in development of the optic vesicle. Melanocytes migrate in the lids before separation, explaining the occurrence of a so-called dividing or kissing nevus. Other lid abnormalities such as epiblepharon, entropion, and ectropion are the result of minor abnormalities in development.

Duke-Elder³ classified the pathology of the eyelids on the basis of chronologic fetal development (Table 34-2).

Tessier^{5,6} classified facial clefts numerically based on their anatomic locations. The clefts are defects resulting from inadequate fusion of soft tissue and bone. The eye, including the eyelids, the orbital cavity, and the brain, reach adult dimensions by 7 to 8 years of age. With the aging process, from middle age into old age, looseness of the skin and ligamentous attachments and dehiscence of the levator aponeurosis are responsible for conditions not generally seen in children. Reconstructive procedures in children, however, use many of the same techniques used in adults.

NORMAL AND ABNORMAL ANATOMY

Eyelids are a highly complex structure composed of an anterior and posterior lamella separated by the orbital septum (Fig. 34-1). Skin and orbicularis oculi form the anterior lamella. The upper lid posterior lamella consists of the conjunctiva, Müller's muscle, levator aponeurosis, and tarsus.

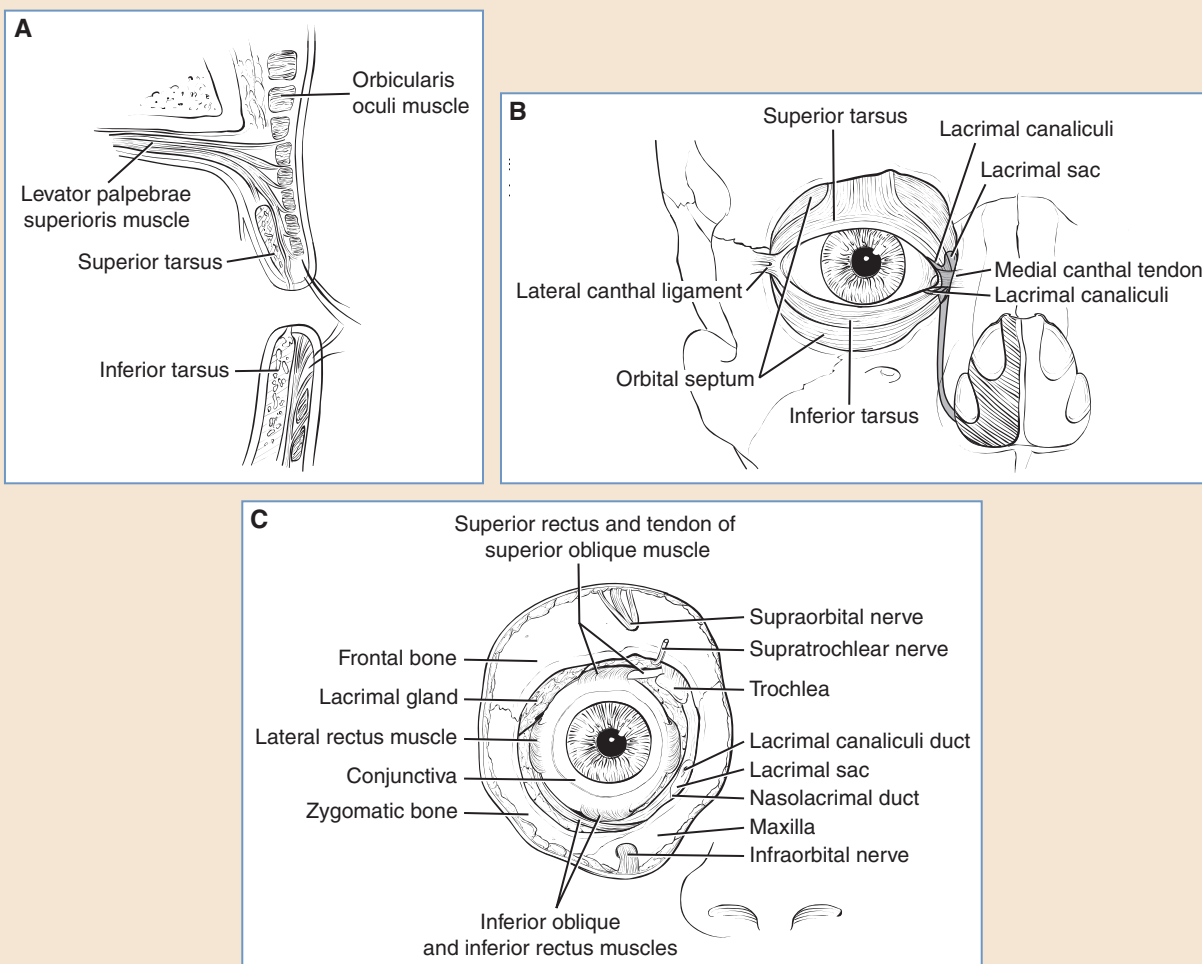


Fig. 34-1 **A**, A lateral view of the eyelid. Upper and lower tarsal plates are evident, with the orbital septum running from the upper portion of the tarsus to the orbital rim and the levator muscle attaching to the levator aponeurosis, which attaches to the tarsus. **B**, A frontal view of the eyelid. The lateral canthal tendon is slightly higher than the medial canthal tendon. The upper and lower puncta pass through canaliculi to the lacrimal sac. The lacrimal conduit passes to its exit point beneath the inferior turbinate. **C**, A deeper view of the orbital structures.

The lower lid posterior lamella is made of the conjunctiva and capsulopalpebral fascia. The upper tarsus is approximately 10 mm high, whereas the lower tarsus is up to 4.5 mm high.⁷⁻⁹

The orbicularis oculi is innervated by the seventh cranial nerve. The eyelid closes on contraction of this muscle. The orbicularis oculi is divided into three muscles: pretarsal, preseptal, and orbital.^{7,8} The levator muscle, innervated by the third cranial nerve, and Müller's muscle, innervated by sympathetics, together raise the upper eyelid. The levator muscle originates at the lesser wing of the sphenoid. It forms the aponeurosis and inserts onto the tarsus.⁸ The lateral attachment ends at the lateral retinaculum, and the medial attachment ends at the posterior lacrimal crest. The distance the upper eyelid moves from downward to upward gaze is a measure of levator function. Müller's muscle, a smooth muscle, originates at the posterior part of the levator and inserts onto the superior part of the tarsus. It is found in the upper lid posterior lamella and is attached to the levator and tarsus. Injury to Müller's muscle results in a 2 to 3 mm ptosis.⁷

The orbital septum prevents the spread of infection and bleeding from the eyelid to the posterior orbital structures. The septum starts at the orbital rim and inserts above the tarsal border into the upper lid and below the tarsal border into the lower lid.^{7,8}

The lateral canthus (or retinaculum) consists of the inferior suspensory ligament (Lockwood's ligament), the levator aponeurotic lateral extension, the pretarsal and preseptal muscle extension, and the lateral rectus check ligament. The lateral canthus inserts in the Whitnall tubercle, located at the lateral orbital wall or the zygoma. This is clinically significant, because inferiorly displaced zygoma fractures cause the palpebral fissure to have a downward slant by pulling down on the lateral canthus. The medial canthus has three insertion points: the anterior lacrimal crest, the nasal bone—close to the nasofrontal suture—and the posterior lacrimal crest. The lacrimal sac is found between the anterior and posterior insertions of the medial canthus.⁷⁻¹⁰

All of the extraocular muscles except the inferior oblique originate near the orbital apex at the annulus of Zinn. The origin of the inferior oblique is the anterior orbit. This muscle separates the lower lid medial and central fat compartments.⁷

The supratarsal fold is the fold of skin above the dermal attachments of the levator aponeurosis and is usually absent in people of Asian descent. This fold is usually 1 cm above the eyelid margin and, if in a high position, the cause may be a levator defect.⁷

The orbit fascial framework comprises extraocular muscle-investing fascia, bulbar fascia, Tenon's fascia, the intermuscular septa, and the check ligaments, which are formed by the medial and lateral rectus muscles. Lockwood's ligament is formed by the intermuscular septum, which is between the inferior rectus and inferior oblique.^{7,10}

Eyelids are important for tear production, because they directly influence the pumping mechanism. Pretarsal muscle causes negative pressure inside the lacrimal sac by pulling on the lacrimal diaphragm. With relaxation, tears enter the nasolacrimal duct. Vertical movement of the eyelids lubricates the eyes and forms a visual axis. Eyelashes filter ultraviolet light and foreign bodies. The medial eyelids contain the canalicular drains. There are several glands along the eyelid margin. Approximately 30 meibomian glands are located within the tarsus of each lid. Apocrine glands of Moll and the pilosebaceous glands of Zeis are found in the distal eyelid margin, anterior to the meibomian glands, which form the lipid part of tears.^{4,8,9}

PREOPERATIVE ASSESSMENT

The highest priority in the management of eyelid deformities is to prevent interference with the normal development of vision.⁴ Therefore the globe must be protected from trauma and desiccation. Before the maturation of vision, prolonged, significant occlusion of the visual axis may result in a deprivation amblyopia. Although many of the conditions that present in this region require evaluation and treatment by multiple specialists, an ophthalmologic consultation should be sought early in the diagnosis and treatment. Full fundoscopic and slit lamp examinations performed with the patient under general anesthesia may be essential.

Few conditions require urgent treatment; these include rapidly enlarging lid and orbital hemangiomas (with risk of either occlusion of the visual axis or late anisometropic amblyopia from deformation of the globe), exposure of the cornea caused by coloboma, and marked exorbitism⁴ (Figs. 34-2 and 34-3). Beyond these conditions that present in the neonatal period, these conditions can occur in infants and even in older children who present to pediatric plastic surgeons. It is critical to understand the initial deformity, the extent of previous treatment, and the duration of the eyelid problem for which the parents are seeking evaluation and/or treatment.

An examination of the eyelids should be systematic, beginning with the upper eyelid. A normal resting lid position is 2 mm below the superior corneoscleral junction. If the lid droops lower, it is considered ptotic. The eyelid should also be evaluated for incomplete closure and retraction. Lesions of the eyelid are assessed for size, location, discoloration, and any associated ulceration

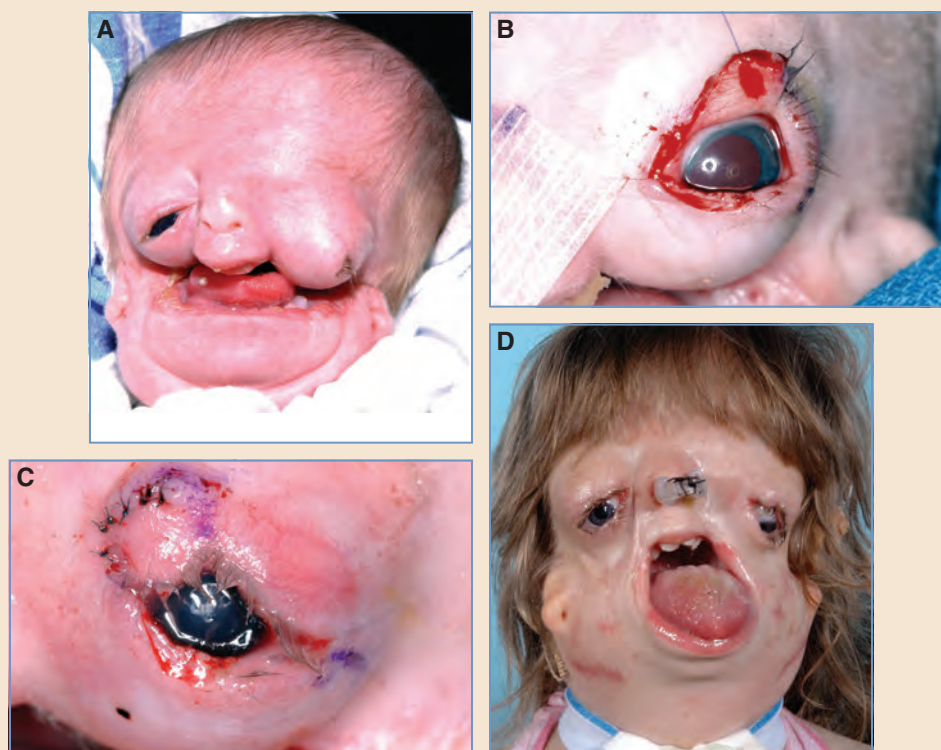


Fig. 34-2 A, This newborn infant was born with a severe form of Treacher Collins–Franceschetti syndrome and was tracheotomized at birth. B and C, The first priority after securing her airway was to correct the left cryptophthalmia, taking the remnant of the lower eyelid and turning it upward to join with the remnant of the upper eyelid. Although the underlying eye is not entirely normal, because it has a coloboma of the iris, the patient has useful vision. D, The patient is shown after a partial correction of her lateral facial clefts, opening of her nasal airways, and correction of an encephalocele. The mandible has been distracted, and a distraction of the LeFort II segment will be performed. (Courtesy of S. Anthony Wolfe, MD.)

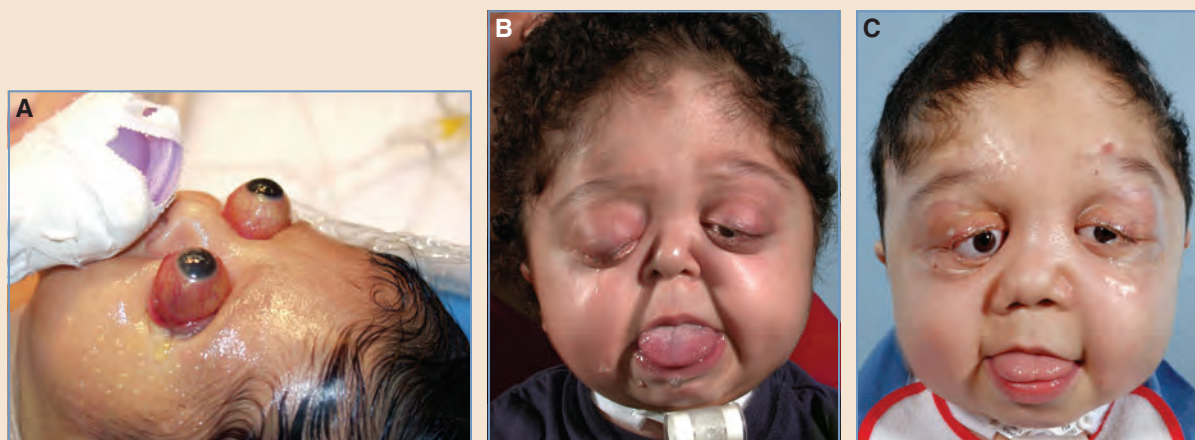


Fig. 34-3 A, This newborn infant had Pfeiffer's syndrome and extreme proptosis resulting from almost nonexistent orbital cavities. B and C, He is shown after two intracranial monobloc frontofacial distractions; the orbital cavities have been developed adequately to allow opening of the tarsorrhaphies. (Courtesy of S. Anthony Wolfe, MD.)

or lash loss. The lower eyelid is inspected for malpositions such as outward turning, referred to as *ectropion*, or inward turning, known as *entropion*. The eyelashes are inspected for misdirection that can irritate the cornea. Some of these deformities and conditions are best treated by a plastic surgeon, whereas others should be treated by a pediatric ophthalmologist. Often, optimal care requires the experience of both specialists.

Pathology of the eyelids can involve soft tissue only or bone and soft tissue. It can arise as an excess, a deficiency, or an aberration from any of the anatomic sites and their components. Although this discussion of defects as separate entities is somewhat arbitrary, it provides a useful framework for discussing many of the congenital presentations. A discussion of the acquired deformities follows. Many of the techniques used in the correction of congenital deformities arose from the reconstruction of acquired defects in adults. Conversely, as combined soft tissue and skeletal reconstruction expanded with Tessier's work and the field of craniofacial reconstruction, the techniques that were designed to correct complex congenital defects were adopted for treating adult eyelid deformities.

Timing of Treatment

The timing of surgical repair of the eyelids depends on the nature of the deformities. Many children with craniofacial syndromes need multiple surgeries arranged in a coordinated fashion. If the globe is becoming desiccated or vision is at risk, eyelid surgery should be performed soon. However, if cosmesis is the main concern, eyelid surgery may be delayed.³

Several important factors require consideration for successful pediatric eyelid surgery. The risk of amblyopia is greater in younger children. Disrupting the binocular vision for even a few days can result in amblyopia. Misalignment of the eyes, occlusive dressings or ointments, and large eyelid masses or flaps can result in vision deprivation and induce amblyopia. The child should be examined by an ophthalmologist if any of these may possibly occur.³

In pediatric patients, the use of absorbable sutures eliminates the need for suture removal. Elbow splints may be necessary to prevent the need for dressing removal and disruption of wounds by the child. To prevent amblyopia, surgeons should not use occlusive dressings or ointments for a prolonged period.⁴

Congenital Deformities of the Eyelid

DEFORMITIES OF THE UPPER EYELID—SOFT TISSUE

Coloboma

A coloboma of the eyelid is defined simply as an eyelid defect. Most occur in the upper eyelid, usually in the medial aspect. They vary in extent from minimal notches to almost complete lack of development of the full-thickness upper eyelid. Colobomas are typically triangular with the base at the lid margin, but defects can be quadrilateral, oval, or irregularly shaped (see Fig. 34-2). Usually, the absence involves all of the lid structures, including skin, orbicularis, tarsus, conjunctiva, lashes, and glands. Partial defects are rare.⁴

The most common position for an eyelid coloboma is at the junction of the medial and middle third of the upper eyelid. Lateral colobomas affecting the lower eyelid are most commonly seen in combination with other soft tissue and skeletal anomalies, as in Treacher Collins syndrome and Goldenhar syndrome^{11,12} (see Table 34-1).

The timing of coloboma repair depends on the degree of corneal exposure caused by the defect. The repair of small or intermediate defects without associated irritation or drying of the cornea may be delayed until the child is 3 to 5 years of age. However, many surgeons prefer to

carry out the reconstruction as soon as the child is a safe anesthetic risk to restore function early and to prevent the possible later stigmata of the deformity as the child approaches school age.

The method of repair depends on the size and location of the defect. Most colobomas are small defects involving less than 30% of the eyelid and can usually be repaired by primary closure. Larger defects, involving 30% to 50% of the lid, will require a lateral canthotomy to obtain adequate relaxation of the lid margin and prevent ptosis caused by a tethering effect on the taut lid. Defects involving more than 50% of the lid margin may necessitate a major lid reconstruction such as a rotation flap created from the lower eyelid.

Defects of the lower eyelid, as seen in the varied oroocular clefts and Treacher Collins syndrome, require a reconstruction of the skeletal and soft tissue components.^{13,14} These will be reviewed in greater detail in the discussion of Tessier clefts, later in this chapter and in Chapters 26 and 30. In the lower lid, three layers need to be reconstructed: the skin, the supporting layer, and the conjunctiva. Skin grafts or a flap may be required to replace the skin. A Mustardé cheek advancement flap is one of several flaps appropriate for filling a large defect of lower lid tissue. The surgeon may need to provide added support (cartilage) or lining (conjunctiva) if more than 30% of the lower lid tissue is missing.^{9,13} Buccal and nasal mucosal grafts can be used to replace conjunctiva, or a septal chondromucosal graft may be used to replace both the intermediate supporting layer and lining as a single graft.

Ptosis

Congenital ptosis is defined as eyelid drooping that presents in the perinatal period. However, recognition of the defect may be delayed because of lid edema in the neonatal period or failure to recognize differences in the two lids when the condition is unilateral.

Congenital ptosis can be sporadic or familial. It is usually unilateral. It may be seen alone or in combination with other extraocular muscle weakness, particularly of the superior rectus muscle.^{4,15,16} Other variants of congenital ptosis include symmetrical ptosis, which is common, and Marcus Gunn ptosis, which is rare. Blepharophimosis is another variant. It involves a decreased palpebral aperture with ptosis, epicanthus, and telecanthus. This can occur as part of a complex of anomalies in blepharophimosis syndrome (Figs. 34-4 through 34-6).

Congenital ptosis seems to arise as a result of a deficiency of striated muscle in the levator palpebrae muscle.¹⁵ This ranges from very mild to near total absence of the muscle (Table 34-3). The severity of muscle deficiency seems to be proportional to the severity of the ptosis and the lack of levator function.

A clinical assessment of ptosis includes the birth history and the family history. The progression of the ptosis, the visual status, and the status of extraocular muscle function should be recorded. Regardless of which surgical discipline will be involved in treating the ptosis, a full examination should ideally be performed by a pediatric ophthalmologist, and visual acuity and extraocular muscle function should be recorded.

Table 34-3 Grades of Ptosis and Levator Function

Ptosis (mm)	Levator Function (mm)
Mild: 2-3	Good: 10-15
Moderate: 3-5	Fair: 6-9
Severe: >5	Poor: <5



Fig. 34-4 **A**, This patient had blepharophimosis with the characteristic telecanthus, epicanthus inversus, ptosis, lack of fixation of the lateral canthus, and short transverse palpebral fissures. **B**, The patient is shown after transnasal medial canthopexies with Z-plasties in the medial canthal area and a lateral canthopexy and a small calvarial bone graft to the nose. The canthopexy wires were tightened over the nasal bone graft through a short incision. (Courtesy of S. Anthony Wolfe, MD.)

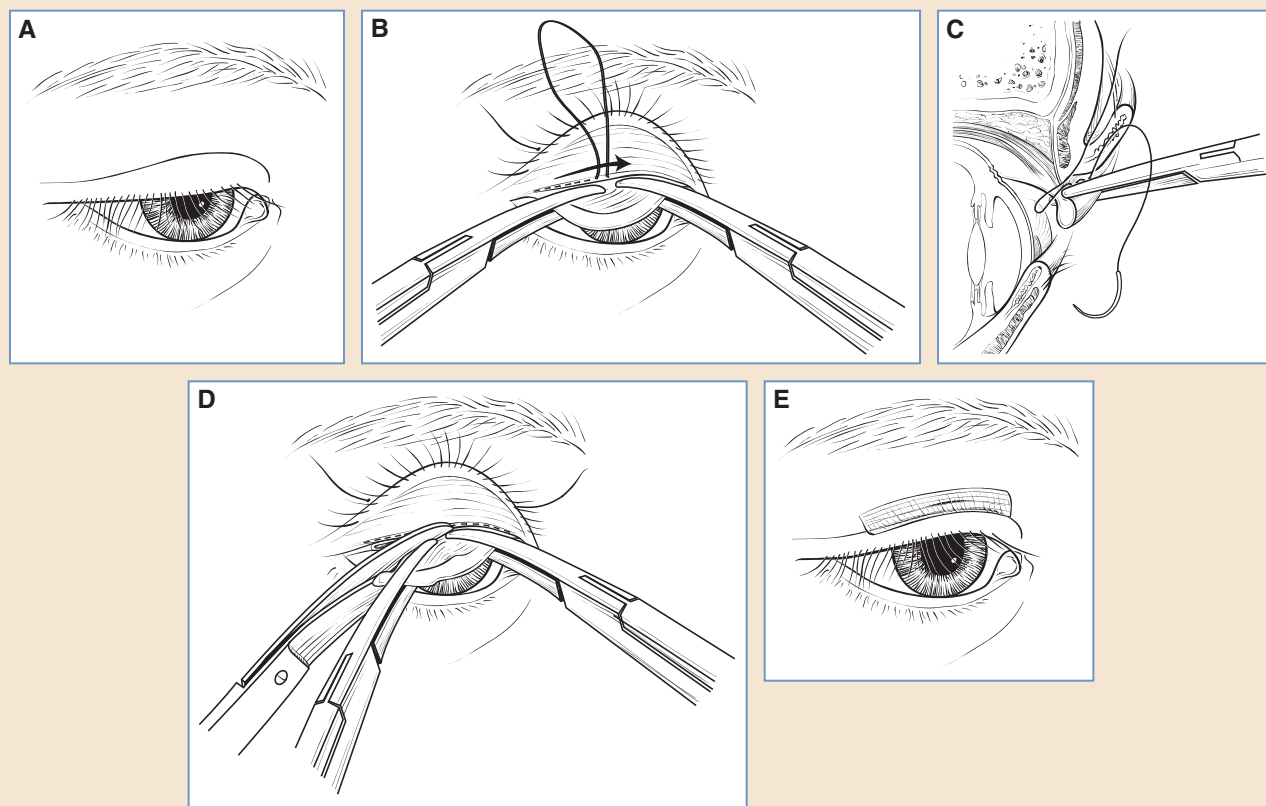


Fig. 34-5 The Fasanella-Servat procedure (tarsconjunctival mullerectomy) is indicated for patients with good levator excursion and mild ptosis. **A**, Mild upper lid ptosis of 1 to 2 mm. **B**, Running suture (full-thickness) is placed behind the clamps from lateral to medial. **C**, A cross-section of the suture placement. **D**, Excess clamped tissue is trimmed. **E**, Closure: Suture is tied on the skin surface and covered with a Steri-Strip bandage.

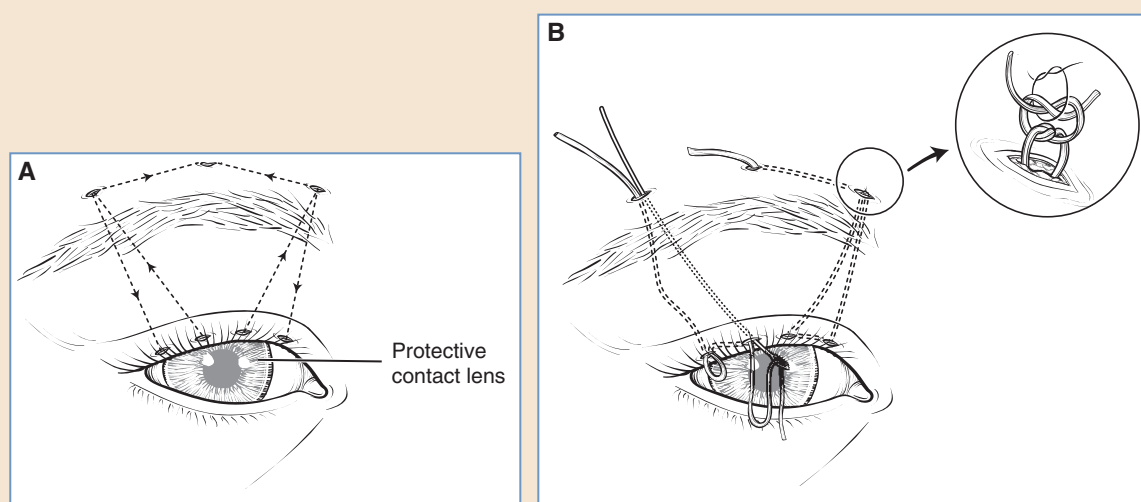


Fig. 34-6 **A**, A frontalis sling tethers the upper eyelid to the frontalis muscle above by way of alloplastic or autogenous material tunneled beneath the orbicularis oculi muscle. A protective contact lens is placed on the eye. **B**, Suspension material is placed subcutaneously to create a static sling. Insert shows a suture loop through the knot tied in the suspension material, allowing later access to the knot and earlier postoperative adjustment of the correction.

The presence of a chin-up head posture should be noted during the examination of a child with congenital ptosis. Such posture can result in neck problems and visual axis occlusion. This may affect ocular motility, levator function, and the location of the eyelid crease. Levator function can be measured by the amplitude of the excursion from downward gaze to maximal upward gaze. If levator function is adequate, the chances for a good repair and adequate lid level after surgery are improved.

The examiner should stabilize the brow with his or her finger to measure the true excursion of the lid. The usual guidelines for measurement and treatment depend on the adequacy of levator function. Finally, the child is tested preoperatively for evidence of Bell's phenomenon. This is a test of the ability of the globe to rotate upward during sleep, an important reflex when inadequate coverage of the cornea is a problem. Its absence indicates a potential postoperative problem if lagophthalmos occurs.

The timing of surgical correction in the absence of significant obstruction of the visual field is often chosen depending on when the parents are motivated and when the child can be adequately evaluated by the surgeon. The child may be about 3 to 4 years of age. Severe ptosis with a significant risk of deprivation amblyopia should be promptly corrected.

Correction of Ptosis

Entire books have been written about ptosis and its correction; therefore it is best to refer to other sources for an in-depth discussion of each of the many techniques, their indications, and the nuances of different approaches. Surgeons who will be treating these patients need to be fa-

miliar with all of the types and degrees of ptosis and able to select and perform the proper ptosis repair for each particular defect.¹⁶⁻²²

The choice of operation is important to ensure optimal correction and symmetry. Generally, these techniques include (1) the Fasanella-Servat procedure¹⁷ (see Fig. 34-5), (2) a resection of Müller's muscle,¹⁶ (3) a levator resection through either an anterior or posterior approach,¹⁸ and (4) a brow suspension with either autologous or alloplastic material¹⁹⁻²³ (see Fig. 34-6).

As stated earlier, the surgical approach depends on the amount of levator function. If good levator function is present (8 to 12 mm), placcation of the levator aponeurosis or excision and repair can be planned. This will usually correct mild to moderate ptosis, and the patient will have little or no lagophthalmos. For congenital ptosis with poor (2 to 4 mm) or fair (5 to 7 mm) levator excursion, a levator shortening or a levator advancement is best. In cases of severe ptosis with minimal or no levator function (less than 2 mm), a frontalis suspension with fascia lata, temporalis fascia, or alloplastic material is the procedure of choice.

A levator aponeurosis advancement is performed by first making a horizontal incision in the lid crease. Scissors are used to dissect through the orbicularis muscle layer and the orbital septum. Once dissection is through the preaponeurotic fat pad, the aponeurotic extension of the levator muscle is identified. This is dissected away from the upper surface of the tarsal plate. After partial resection, the aponeurosis is advanced and sutured to the upper tarsal plate. For each millimeter of ptosis, the levator aponeurosis should be resected 4 mm. As a guide, one plus the amount of preoperative lagophthalmos added to the degree of blepharoptosis should equal the final amount of lagophthalmos in millimeters.²⁴

To perform a Whitnall's sling, after identifying the aponeurosis, Whitnall's ligament in the superior orbit is sutured to the tarsal plate. Nonabsorbable sutures are used. The other approach is to transect the levator muscle just above Whitnall's ligament, advance it over the levator aponeurosis, and suture it to the tarsal plate.^{20,24}

A frontalis suspension elevates the brow by using a sling material to connect the frontalis muscle superior to the brow to the eyelid tarsus. Types of materials used are Gore-Tex,²³ silicone rods, polypropylene suture, and allogeneic fascia lata or autogenous fascia lata.²² Autogenous fascia lata has been reported to have the lowest rate of infection.³ Although some prefer to use alloplastic materials, autogenous tissue even in younger patients can be readily harvested (see Fig. 34-6).

Epicanthus

Epicanthal folds are vertical skin folds in the area of the medial canthus. The skin arches along the side of the nose, with the concavity directed laterally. Most arise from the upper lid above the tarsal region and extend to the lower orbital margin; however, variations occur.^{4,24,25} Epicanthal folds are categorized in four groups: (1) palpebralis, (2) tarsalis, (3) inversus, and (4) supraciliaris. *Epicanthus palpebralis* involves an even distribution of the fold between the upper and lower lids. *Epicanthus tarsalis* refers to the skin fold being greatest in the upper lid. In *epicanthus inversus*, the fold is greatest in the lower eyelid. In *epicanthus supraciliaris*, the fold is between the eyebrow and the lacrimal sac. Congenital epicanthus is most commonly seen in Asians.²⁴ Epicanthal folds may be seen in isolation or associated with syndromes, usually blepharophimosis syndrome (see Fig. 34-4). Although this topic is somewhat arbitrarily separated as a disorder of the soft tissue of the upper lid, cases seen in association with telecanthus and with those having tissue shortages in both the upper and lower lid are reviewed in the discussion of medial canthal deformities.

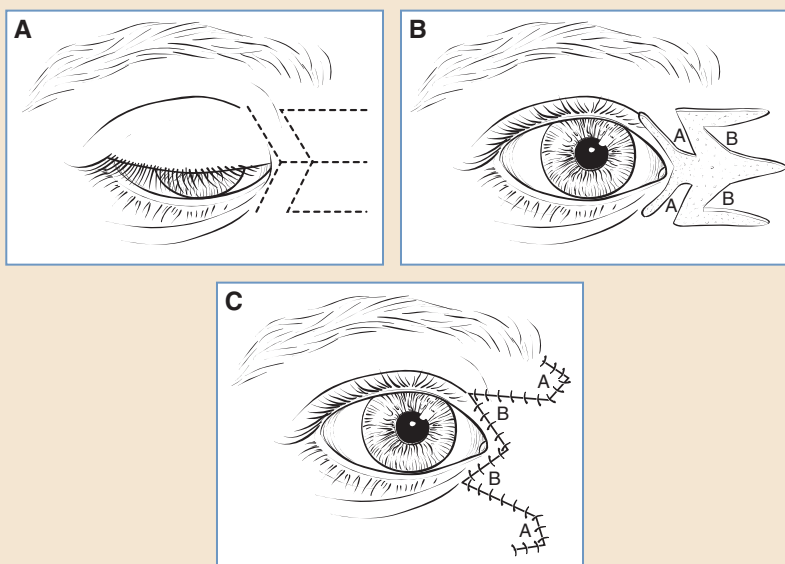


Fig. 34-7 The Mustardé four-flap technique to correct epicanthal folds. **A**, A mark is made midway between the nasal midline and the center of the pupil. Medial traction is placed on the nose. The medial canthus is marked, and a line is drawn between these points. At the center of this line, two lines are drawn at 60 degrees approximately 1 to 2 mm shorter than the line. Two lines are drawn at 45 degrees, and two paramarginal lines are marked. **B**, The incisions are made, and the flaps are raised. **C**, After the transposition is completed, the flaps are sutured.

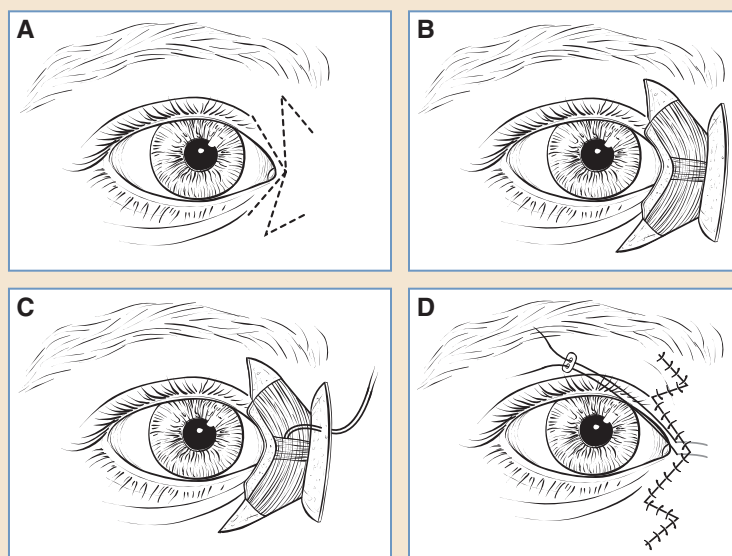


Fig. 34-8 Double-opposing Z-plasties for the correction of traumatic epicanthus. **A**, The site is marked. **B**, Incisions are made, and flaps are raised. **C** and **D**, Transosseous wires are secured through the nasal bones, and the flaps are transposed and sutured. Pledgets are tied in place.

Correction of Epicanthus

Epicanthal folds usually resolve with normal development, especially the supraciliaris or palpebralis types. Simple, nonresolving epicanthus is corrected with Z-plasties, one above the canthal angle and another below it. The Mustardé four-flap technique is effective for epicanthus inversus, characterized by an abnormal lower lid fold¹³ (Fig. 34-7). Many surgeons have found that the multiple Z-plasty scars in this region leave residual skin irregularities that make the scar quite visible even though the contour of the canthal area is more normal. For this reason, repairs with fewer Z-plasties than the typical Mustardé four-flap procedure may be preferred in many cases and may result in excellent correction and less visible scars.

Traumatic epicanthus can be corrected with double-opposing Z-plasties (Fig. 34-8). This technique releases the tension and the scar.^{4,7,24} Z-plasties are made opposite each other. After the flaps are raised, the scar tissue is removed. Transosseous wires are placed through the nasal bones. Wires are placed through and through, and the canthal buttons are placed.¹³

DEFORMITIES OF THE LOWER EYELID—SOFT TISSUE**Epiblepharon**

Epiblepharon is characterized by eyelashes that are vertical, because pretarsal muscle and skin overrides the margin of the eyelid, often affecting the lower lids. The lower eyelid may have a congenital absence or deficiency of the adhesion between the lower eyelid retractors and the anterior lamella that allows the skin and muscle to roll upward, accompanied by a hypertrophied orbicularis with an extra skin fold. This condition more often affects Asians and usually resolves spontaneously in the first few years of life.²⁵

Correction of Epiblepharon

Surgical treatment is indicated if epiblepharon does not spontaneously resolve or if the cornea becomes irritated. Repair involves shortening the anterior lid lamella, which is accomplished by excising a horizontal piece of skin with orbicularis muscle. The skin edges are sutured to the eyelid retractors or anterior tarsal surface using absorbable sutures. The deformity can also be corrected by resecting part of the medial canthal tendon and using the Mustardé four-flap technique.^{3,11} When repositioning of the canthal ligament is required, transnasal wires are passed through the end of the medial canthal tendons, and buttons are placed.

Epiblepharon can occur after trauma, burns, and fractures. In these cases, scar tissue may need to be excised, and the inferior part of the lateral canthal tendon may need to be cut to release the tension before transnasal medial canthoplasty.¹¹

Ectropion

Congenital ectropion and oversized eyelids are associated with other lid deformities such as distichiasis and with general congenital abnormalities such as trisomy 21.⁴ Congenital ectropion as an isolated occurrence is unusual and may be confused with euryblepharon. The degree of lower lid soft tissue deficiency varies, as does the degree of exposure of the globe (Figs. 34-9 and 34-10). The tarsal plate, along with the lateral canthal tendon, may be hypoplastic and give little purchase in efforts to use this structure for support. Full-thickness reconstruction of the lower eyelid may be required if the ectropion is severe and cannot be treated by the usual methods of tarsal tightening or skin grafting.

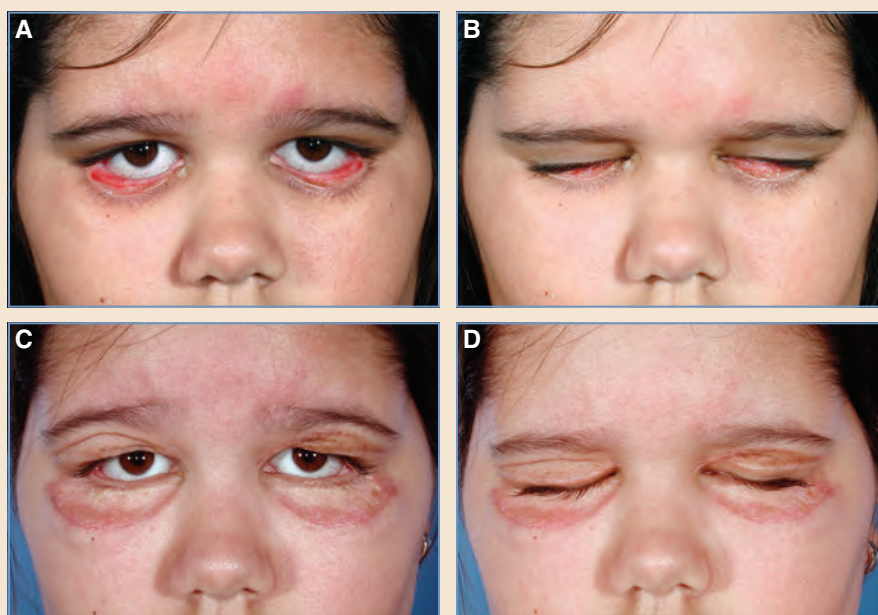


Fig. 34-9 A and B, This 9-year-old girl had congenital ectropion of her upper and lower eyelids. C and D, She is shown after full-thickness postauricular skin grafting to the lower lids and cheeks and a medium-thickness split graft from the upper inner arm to the upper eyelids. (Courtesy of S. Anthony Wolfe, MD.)

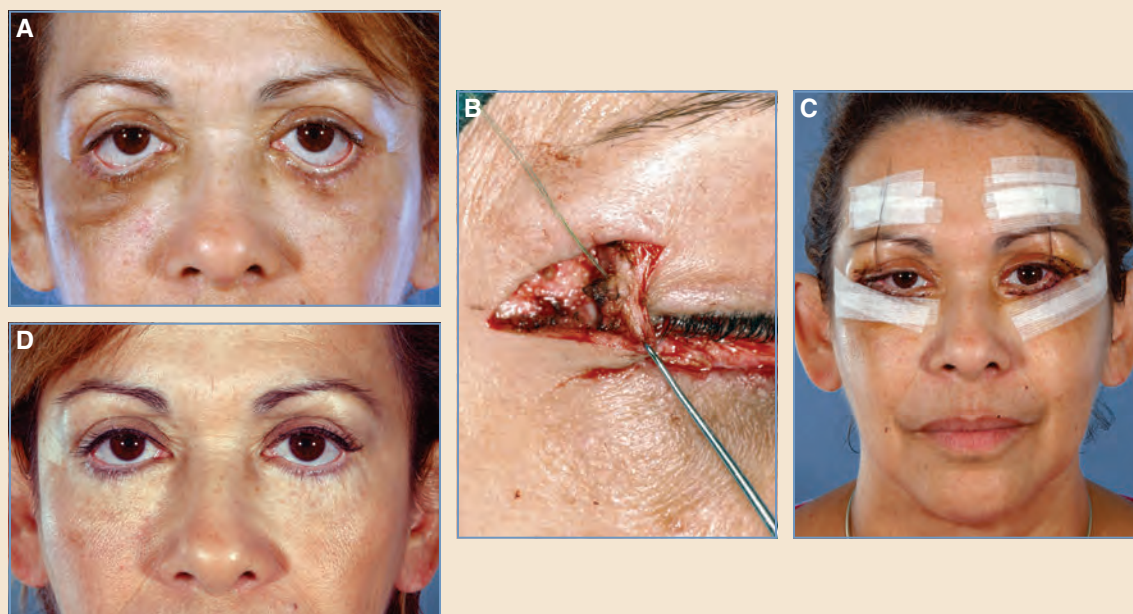


Fig. 34-10 Secondary ectropion presents with similar problems of tissue shortages and reconstructive needs in many congenital cases. A, This patient presented with significant bilateral ectropion after a midface lift through the lower eyelids and a second attempt to correct them elsewhere. B and C, Correction entailed releasing the lower lids completely through the previous subciliary incision, excising scar tissue, performing a firm lateral canthopexy with a dermoorbicular pennant flap brought through the lateral orbital wall, and the use of Frost sutures for 1 week and then taping the eyelids for another month. D, The postoperative result without further skin grafting. (Courtesy of S. Anthony Wolfe, MD.)

Correction of Ectropion

Various wedge excisions have been suggested for the treatment of severe ectropion. Tarsorrhaphy sutures are placed to provide corneal protection. Markings are made below the ciliary margins and infiltrated with saline solution and epinephrine. A skin incision over the orbicularis muscle is made, and about a 7 to 8 mm strip of lid margin is deepithelialized and a tarsal strip produced. An incision is made in the upper lid, just beneath the brow in the position where the tarsal crease will eventually be. The periosteum and superior lateral orbital rim are dissected, and a scissor is passed from the supraperiosteal plane down to the tarsal strip area. The conjunctiva and lid margin can be closed with 5-0 plain catgut. A double-armed 4-0 Vicryl suture is taken through the tarsal strip and sutured to the inner lip of the periosteum of the orbital rim (see Fig. 34-10). Tarsorrhaphy sutures are taken between the upper and lower lids. The upper lid incision can be closed with 7-0 silk. The bipediced orbicularis oculi muscle flap is freed and sutured over the tarsal plate with 7-0 Vicryl. A postauricular skin graft is harvested and sutured in place with 7-0 silk, and a tie-over bolus of antibiotic ointment is placed with 6-0 silk. The donor area is closed with 4-0 Monocryl.

Entropion

Congenital entropion is a rare condition in which the eyelid margin is turned inward, and the eyelashes are oriented toward the cornea. It can affect the entire lid. The conjunctiva may become irritated and cause itching, with resultant rubbing of the eyelid. It is theorized that this action causes chronic inflammation and swelling of the orbicularis muscle and overlying skin and soft tissue, accounting for persistent congenital entropion. This frequently results in orbicularis oculi spasm.^{4,24}

Epiblepharon differs from entropion²⁵ in that occasionally the fold of skin in the medial lid may cause some of the lashes to irritate the cornea; for epiblepharon, simple resection of the medial fold of skin will usually cure the problem. An additional difference between congenital entropion and epiblepharon is the positioning of the lid margin in relation to the globe, with the rarer entropion causing more difficulties with trichiasis because of malpositioning of the lower eyelashes.^{26,27}

Ito et al²⁸ described correction of entropion with a horizontal lower eyelid incision below the eyelashes. The pretarsal orbicularis muscle is mobilized with resected excess muscle and skin. If the entropion is more severe, the attached skin is expanded and sutured at the tarsal plate. Once the position of the eyelashes is acceptable, the skin incision is sutured closed.

Distichiasis

Distichiasis refers to an accessory row of lashes along the posterior marginal edges of the lid, occupying the site of the meibomian glands (which they often replace). It varies from trichiasis, in which irregularly placed cilia associated with entropion turn toward the cornea after injury or disease.⁴

Surgical correction involves excising approximately 4 mm of the posterior lamella of the tarsus and conjunctiva, including the posterior row of lashes from the edge of the lid; excision should include all of the abnormally placed follicles. The mucosal defect is grafted with nasal or buccal mucosa. This is preferable to excision and direct closure by tarsoconjunctival advancement, which may result in entropion and recurrent trichiasis. Another procedure involving cryotherapy of the posterior eyelid margin may be performed to destroy these extra lash follicles.⁴

Euryblepharon

Euryblepharon is a widening of the palpebral fissure laterally and vertically caused by a shortage of eyelid tissue. Vertical widening is most prominent laterally. The lateral canthal tendon may be displaced inferiorly, resulting in an antimongoloid slant. Usually, a gutter has formed between the lower lid and the eye.^{4,24}

Topical lubricants are indicated for the management of corneal exposure. If unsuccessful, then surgical repair is necessary. Congenital ectropion treatment, as for acquired ectropion, may need to include full-thickness skin reconstruction, a lateral canthopexy, and a tarsal strip procedure or full-thickness lid reconstruction.^{26,28}

Euryblepharon is sometimes seen in patients with Apert syndrome. It may be improved by advancement of the bony orbit and reduction of the prominence of the globe to the extent that it is less likely to push the eyelids to a wide open position.

Ankyloblepharon and Cryptophthalmos

Ankyloblepharon is fusion or incomplete separation of the upper and lower eyelids. This condition may be inherited in an autosomal dominant manner. Fusion can be lateral or medial.²⁴ Ankyloblepharon filiform adnatum consists of many tissue strands between the upper and lower eyelids. This may be associated with central nervous system, cardiac, and ectodermal findings.²⁹

The treatment of ankyloblepharon is surgical. The eyelids are separated, with careful attention to the medial aspect to identify the lacrimal ducts.³

Cryptophthalmos is a failure of normal embryonic development of the lid fold (epithelium), which normally differentiates into the cornea and conjunctiva. The lid folds fail to develop, and the eye is buried in the developing cover of the epithelium but does not differentiate normally. Cryptophthalmos is inherited as an autosomal recessive congenital defect. It may be associated with syndactyly and other congenital defects, including cardiac, facial, ear, and cortical developmental defects (Fig. 34-11).

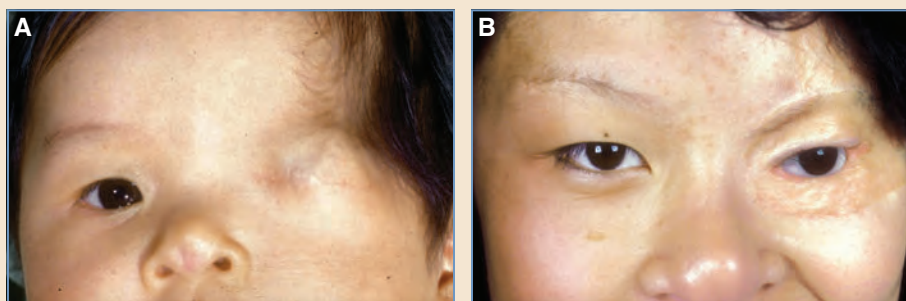


Fig. 34-11 A, This patient had congenital cryptophthalmia. She is shown before reconstruction of the eyebrow and full-thickness skin grafting to the upper and lower lids and eyelash grafting to the upper eyelids and brow. An ocular prosthesis was placed. B, The postoperative result. (Courtesy of Paul Tessier, MD.)

Because the eye does not differentiate normally, surgery will not successfully expose and preserve the globe.

MEDIAL CANTHAL DEFORMITIES

Medial Canthal Dystopia

Malposition of the medial canthal tendon is rarely an isolated finding, but it occurs with naso-ocular clefting. To correct the downward displacement of the medial canthal tendon, the tendon must be freed of all periosteal attachments and replaced upward to attach to the lacrimal bone in a more normal position. This reconstruction may best be done in concert with additional craniofacial procedures performed to treat the complex skeletal and soft tissue deformities inherent in the surface and underlying bony clefts.⁴

Telecanthus and Blepharophimosis Syndrome

Telecanthus alone is known as *Waardenburg syndrome*.⁴ Telecanthus in association with epicanthus inversus, decreased horizontal lid fissure, and ptosis is known as *congenital tetrad syndrome* or *blepharophimosis syndrome* and is an autosomal dominant disorder. In blepharophimosis syndrome, the eyebrows appear arched from the ptosis.²²⁻²⁴ Other findings include ectropion of the lower lid, flattening of the nasal bridge, and hypoplasia of the superior orbital rim. Usually, the supratarsal fold of the upper lid is absent. Forehead and/or ear deformities may be present. The neck may be extended, allowing the patient to see below his or her ptotic eyelids.²⁴ Patients with type I blepharophimosis have blepharophimosis with epicanthus inversus and ptosis. The findings in type II include telecanthus, ptosis, ectropion of the lower lids, absent epicanthal folds, and an insufficient amount of skin in all lids. Type III is characterized by telecanthus, ptosis, hypertelorism, slanting palpebral fissures, and an insufficient amount of eyelid skin.¹³

Correction of Telecanthus and Blepharophimosis Syndrome

Surgical correction is often performed by the time a patient is 3 to 5 years of age, after the nose and midface are able to grow.²²⁻²⁴ For those with type I blepharophimosis, the quadrilateral flap technique described by Mustardé or a Z-plasty (double-opposing) is used.¹³ A bony window can be created through the lacrimal groove. Medial canthal tendons are transnasally wired and advanced into the window (Fig. 34-12). The severe ptosis is often corrected by suspension of the frontalis muscle. Type II correction involves correcting the deficient skin with skin grafts. Medial canthal tendons are fixated into the medial orbits and by transnasal wiring (see Figs. 34-4 and 34-12). Other modes of treatment may involve medial translocation of the medial orbital walls and transnasal wiring of the medial canthi. Ptosis is corrected by frontalis suspension or other techniques. Correction of type III involves skin grafting of the eyelids and medial canthoplasty. Hypertelorism is corrected by moving the orbits medially. Ptosis is also corrected in this manner.^{13,19,22,23}

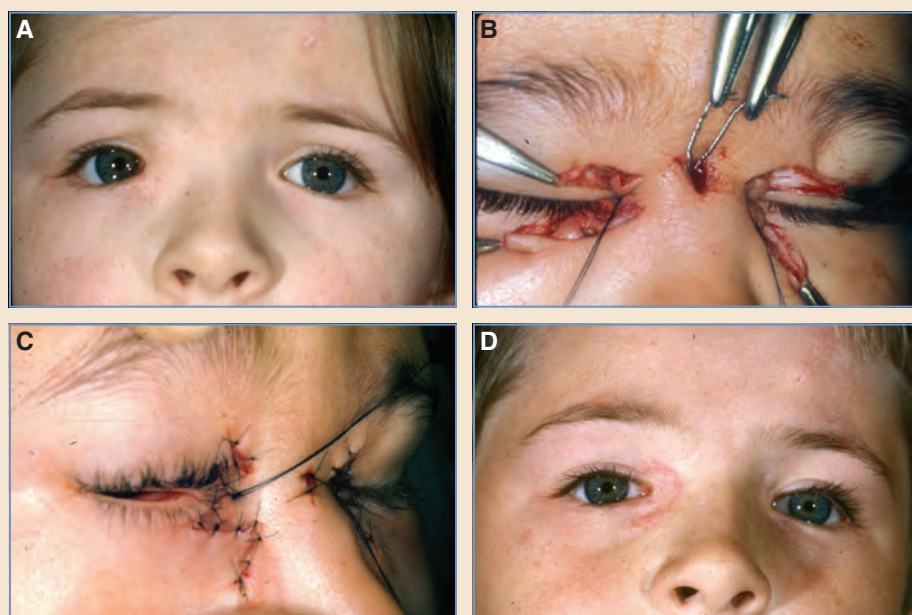


Fig. 34-12 A, This patient had blepharophimosis. B and C, Treatment involved a transnasal medial canthopexy and Z-plasties over the epicanthal folds. D, The postoperative result. (Courtesy of Paul Tessier, MD.)

FACIAL CLEFTS—COMBINED BONE AND SOFT TISSUE DEFECTS

Although combined skeletal and soft tissue eyelid defects are discussed in Chapter 26, we will present some additional defects in this chapter. As noted previously, colobomas of the medial and lateral lower eyelid and medial upper eyelid can be associated with incomplete facial clefts. They manifest mostly as a soft tissue deformity; however, in general, they are associated with an underlying bony cleft.^{5,6,30-34}

Tessier Classification

In 1976 Paul Tessier⁵ published his classification of facial clefts.⁶ He designed this to be an orbitocentric classification, with the clefts passing like time zones through the orbit. Clefts that involve the lower eyelid include Tessier 3, 4, 5, and 6 (Figs. 34-13 through 34-18). In addition to his enormous contributions—which have resulted in the new field of craniofacial surgery—and the Tessier cleft classification, Paul Tessier was an orbital and eyelid surgeon without equal. The literature includes many examples of complex and sophisticated bony reconstructive procedures in the periorbital area that left residual soft tissue deformities uncorrected, allowing the stigmata of the defect to persist. Tessier's cases, illustrated in the following section, clearly demonstrate his results as the benchmark. It is highly unlikely that another surgeon will have his vision, experience, and technical ability.

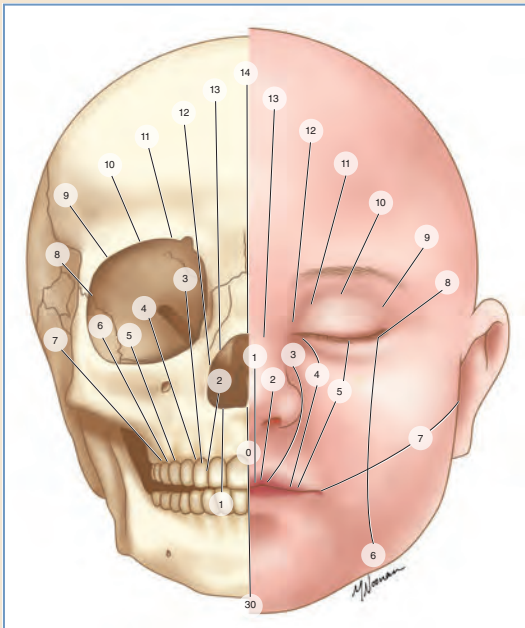


Fig. 34-13 Tessier's classification of facial clefts, designed as an orbi-centric classification with the clefts passing like time zones through the orbit, showing underlying skeletal defects and soft tissue clefts.



Fig. 34-14 A and B, Bilateral Tessier number 3 oroocular clefts in full-face and close-up views at birth. C and D, Similar views at 8 years of age. (Courtesy of S. Anthony Wolfe, MD.)

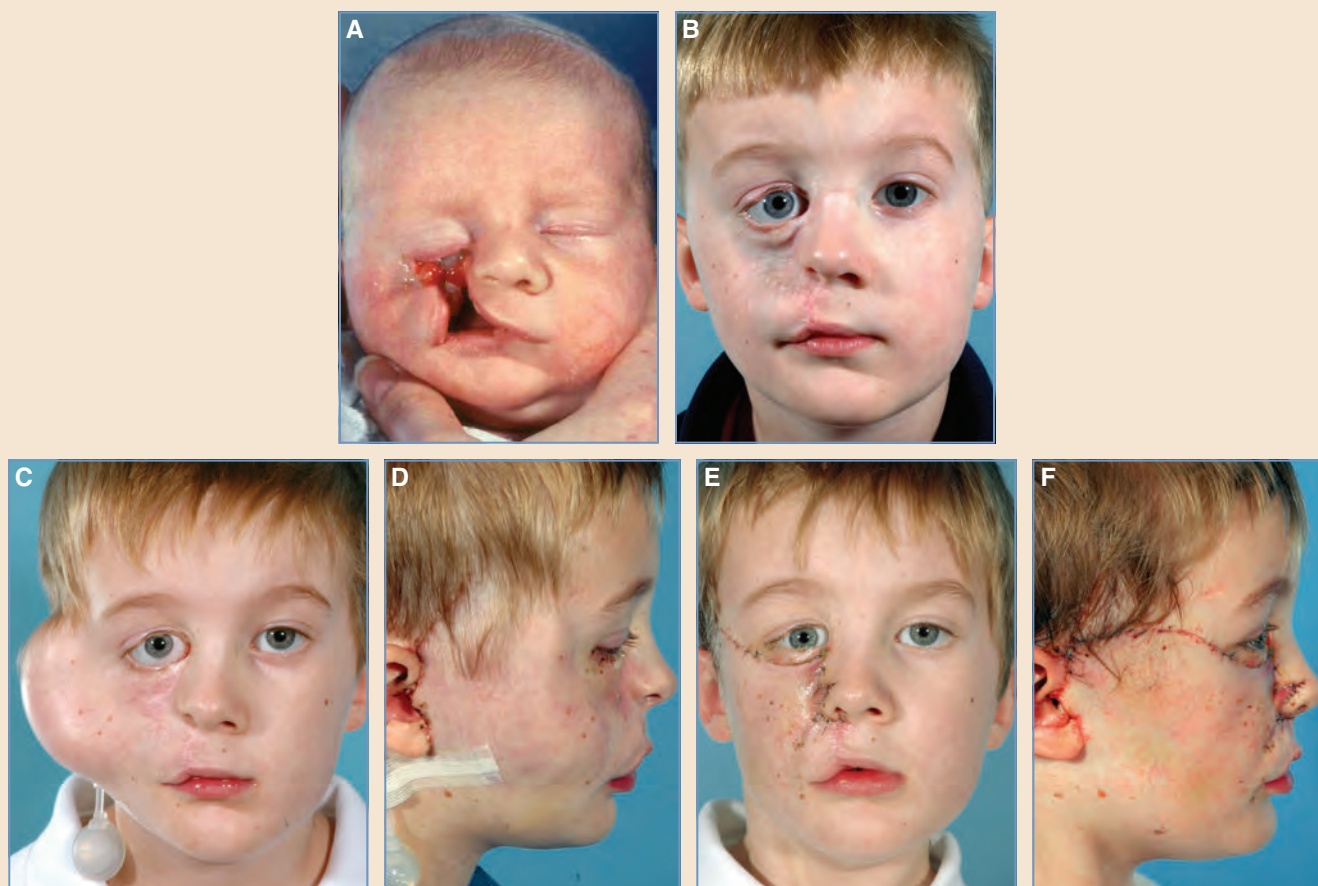


Fig. 34-15 This child had a complete Tessier number 4 cleft and was treated at 5 months of age with lower eyelid construction, a cleft lip repair, and a primary cranial bone graft to the orbital floor. **A**, The appearance of the cleft at birth. **B**, The appearance at 5 years of age. **C** and **D**, The child is shown during the course of tissue expansion at 7 years of age. **E** and **F**, Early postoperative results. (Courtesy of S. Anthony Wolfe, MD.)



Fig. 34-16 **A**, Bilateral Tessier number 4 clefts before treatment. **B**, Eight years after repair. (Courtesy of S. Anthony Wolfe, MD.)



Fig. 34-17 A and B, Bilateral Tessier number 3 oroocular clefts in full-face and close-up views at birth. C and D, Similar views at 8 years of age. (Courtesy of S. Anthony Wolfe, MD.)

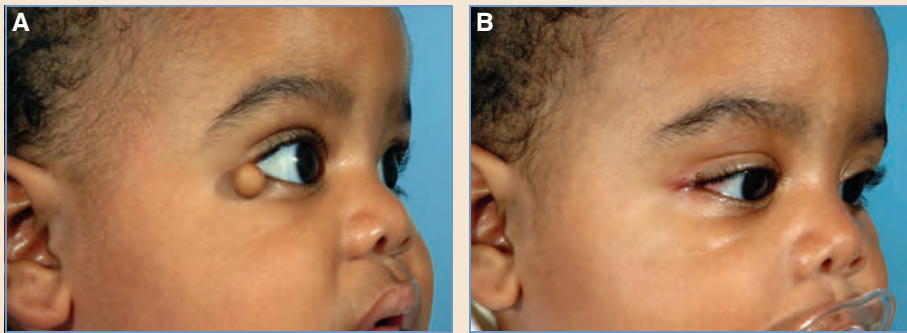


Fig. 34-18 A, This child was born with a soft tissue mass growing from the sclera in association with a Tessier number 6 cleft. B, Postoperative appearance. (Courtesy of S. Anthony Wolfe, MD.)



Fig. 34-19 A and B, This 8-year-old girl had Treacher Collins–Franceschetti syndrome. C and D, The patient is shown after reconstruction of the zygomatic arches with cranial bone, lateral canthopexies, and, at a later stage, a fat dermal graft to the malar area and a genioplasty. (Courtesy of S. Anthony Wolfe, MD.)

Treacher Collins syndrome, also known as *Franceschetti complex*, is a maxillary-zygomatic cleft with a coloboma of the lower eyelid, between the middle and lateral thirds. The cleft is between the maxilla and zygomatic bone, opening into the infraorbital fissure. The cheek area has a further furrow or cleft angling downward. This is a Tessier number 6 cleft. The Tessier number 7 cleft is a temporozygomatic cleft with absence of the zygomatic arch and deformities of the mandible (Fig. 34-19; see Fig. 34-2). The sporadic form of this defect is seen in hemifacial microsomia and in varying asymmetrical degrees in Goldenhar syndrome. In the lateral views of this patient, both ear anomalies and soft tissue deficiency are visible. The deformities are discussed in detail in Chapter 30.

Amniotic Bands

Ocular and oroocular clefts related to amniotic band disruption during facial development present with all of the complexity of soft tissue and bony deformity seen in the group of Tessier clefts discussed previously, but they do not fit into the traditional cleft trajectories. These can be seen alone or in combination with more typical clefts because of the disrupting effect of the encircling amniotic band on the normal developmental processes.³⁵ Fig. 34-20 shows a patient with amniotic bands, a cleft lip, and right microphthalmos before and after multiple reconstructive procedures, including transcranial elevation of the left orbit, reconstruction of the right orbit and right socket, nasal bone grafting, a LeFort type I osteotomy, and a sagittal split of the mandible. Despite multiple procedures, these reconstructions can fall short of an ideal outcome.

Socket Deformities—Combined Bone and Soft Tissue

Microblepharon and Ablepharon

Development and growth of the globe is the primary stimulant for growth and development of the bony orbit. If the eye does not develop, the bony orbit will not develop, resulting in asymmetry of the orbits and possibly soft tissue hypoplasia. In these cases of microblepharon or ablepharon, the usefulness of the globe or remnant must be evaluated before beginning treatment.



Fig. 34-20 A, This child was born with oroocular clefting caused by amniotic band disruption, a cleft lip, and right microphthalmos. B-E, The patient is seen at 1, 2, and 19 years of age, before and after multiple reconstructive procedures, including transcranial elevation of the orbit, reconstruction of a right orbit and right socket, a nasal bone graft, a LeFort type I osteotomy, and a sagittal split of the mandible. (Courtesy of S. Anthony Wolfe, MD.)

Microblepharon is a rare, full-thickness shortening of eyelid tissue often with absent eyelashes and eyebrows and nearly always with severe hypoplasia of the globe. It is usually unilateral. Management is determined by the visual acuity of the small eye. Ablepharon is more extreme, with aplasia of the globe.^{4,29}

Anophthalmos and Microphthalmos

Clinical anophthalmos is the term used by Duke-Elder³ to denote most of the cases in which the eye appears to be absent, although such a socket may contain an extremely small microphthalmic eye (see Fig. 34-11). The lids are variably hypoplastic with a short palpebral fissure and small socket.^{4,36}

The following four factors are critical in the treatment of the hypoplastic orbit and lids:

1. If the orbit is hypoplastic, expansion with progressively larger, expanding orbital implants must be carried out and continued through the period of normal orbital growth.
2. Expansion is more easily obtained by starting earlier than by starting later.

3. The soft tissue of the orbit often requires reconstruction to facilitate the use of an ocular prosthesis.
4. The decision to remove the remains of a poorly developed eye should be made by an ophthalmologist because of other factors related to the globe.

TUMORS AND CYSTS

Dermoid Cysts

Dermoid cysts are hamartomas consisting of ectodermal elements. They typically present as collagenous tissue containing hair follicles and sebaceous and sweat glands and are usually covered with keratinized epithelium. In most cases, dermoid cysts are fixed to the orbital rim periosteum at the frontozygomatic suture (so-called external angular dermoids) but can be loosely attached, allowing them to drift down onto the lid (Fig. 34-21). They may sit in a deep trough in the orbital rim. Rarely, they are intraosseous (see Fig. 34-21, *F*). With the exception of rare intraosseous cysts (which may erode intracranially), they do not extend intracranially.³⁷ It is not clear why this pattern is so common, but it is of interest that they lie along the trajectory of the Tessier number 10 cleft. They may also present more medially along the brow and supraorbital rim. Once they approach the frontonasal junction, they should be evaluated in the same way as a frontonasal dermoid (which may follow well-described paths of intracranial extension).

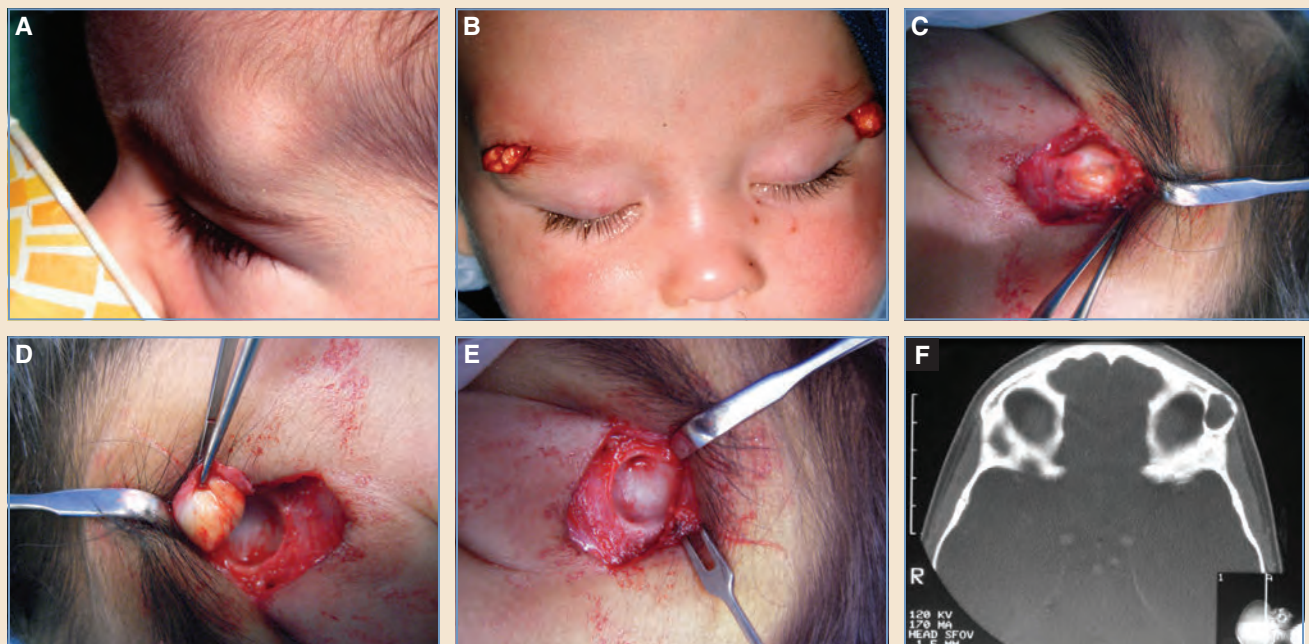


Fig. 34-21 A, A typical position for an external angular dermoid cyst. B, Unusual bilateral cysts before excision. C-E, The cyst in this patient contains well-developed hairs. On excision, the trough in the underlying bone is visible. F, An axial CT view of another patient shows a rare intraosseous dermoid in the bone of the left lateral orbit. (Courtesy of S. Anthony Wolfe, MD.)

Dermoid cysts may grow slowly and rarely reach 4 cm in size. Those in closer contact with the bone may deform it further as they expand. They can become inflamed and rarely infected. Some fluctuate in size, and this may relate to varying activity of the sweat glands present. From a timing standpoint, little is gained in waiting for the cysts to potentially grow larger, and excision any time after a patient has reached 6 months of age, when the anesthesia risk is lower, makes these cases very uncomplicated.³⁷

Most external angular dermoids (see Figs. 34-21, *A* through *E*) can best be excised through an incision in a lid fold of the upper outer eyelid. Dermoids always lie deep to the orbicularis muscle. Those that drift higher toward the temporal region (more than half the distance to the hairline) can be approached through a hairline incision. Endoscopic-assisted excision has been suggested, but this has been associated with injury to the temporal branch of the facial nerve and is therefore discouraged.

Hemangiomas

Hemangiomas are benign vascular tumors with a triphasic growth cycle: proliferation, cessation of growth, and involution. They may present as superficial “strawberry” lesions, deep lesions within the subcutaneous tissue and orbit, or a combination of both. They can be present at birth or develop postnatally, when a small, reddish lesion (herald spot) expands into a much larger growth. Although many hemangiomas will involute without residual tissue, those typically seen by plastic surgeons tend to result in some degree of residual scarring, subcutaneous fibrofatty excess, and associated lid laxity^{4,38} (Figs. 34-22 and 34-23).

Hemangiomas of the eyelid (particularly the upper eyelid) and orbit present one of the true emergencies in the treatment of orbital lesions.^{4,38-40} Early treatment is directed toward preserving vision and preventing the associated amblyopia seen with bulky hemangiomas of the eyelids.⁴⁰ Treatment may involve systemic steroids alone, intralesional injection of combined fast- and slow-acting corticosteroids, or, very rarely, surgery. Some authors report the most effective treatment to be intralesional injection of steroids.²⁴ A combination of short- and long-acting corticosteroids such as betamethasone 0.5 to 1 mg/kg and triamcinolone 3 to 5 mg/kg can be injected with the greatest result at 2 to 4 weeks. Repeat injections are usually needed at 4 to 8 weeks.^{38,39} Although rare, complications of such therapy include deposteroid embolization from injection into the arterial channels of the hemangioma, Cushing syndrome, adrenal suppression, eyelid necrosis, and fat atrophy. Before and after treatment, surgeons should evaluate the retinal vessels for central retinal artery occlusion.⁴¹ Historically, systemic steroids, alpha interferon, and vincristine were used for treating rapid, uncontrolled growth of a hemangioma. Propranolol is currently the drug of choice for hemangiomas with rapid, uncontrolled growth and those whose growth places critical structures at risk, for example, the nose, lips, or ears.

Residual surface vascular changes, scarring, and excess fibrofatty tissue may distort the eyelid and brow. Procedures should be staged to improve contour and correct ptosis of the upper lid and to prevent overexcision that may result in exposure problems and ectropion of the lower lid (see Figs. 34-22 and 34-23). Procedures may be spaced over many years. They typically decrease in the degree of complexity, but minor revisions may be needed in a patient's adult years as age-related changes affect the normal areas and areas previously involved by the hemangioma. When residual vascular changes are noted in skin that cannot be resected without the risk of lagophthalmos or ectropion, the use of a tunable dye laser should be considered.^{4,38}



Fig. 34-22 A and B, This 3-year-old was treated with oral steroids and no other intervention during the proliferative phase of her hemangioma. Despite the size and obstructive nature she was spared visual problems. With constant manual lifting of the hemangioma she could see. C, At 3½ years of age, she had debulking with limited skin resection above the lid fold and in the medial canthal area, with partial correction of ptosis and definition of her lid fold. D and E, She is shown at 4 years of age, before and immediately after her second-stage debulking with limited skin excision. F, Immediately after a third-stage reduction at 4½ years of age. G, At 6 years of age. H, At 8 years of age, 2 years after the stage-three reduction, only fine telangiectatic vessels are visible and lid symmetry is good.



Fig. 34-23 A, At 11 months of age, this girl had extensive involvement of her face despite early systemic steroids and injection of the upper eyelid with combined rapid- and slow-acting steroids. B, At 31 months of age, improvement was substantial. C, At 4 years of age, after the first debulking. D, At 5½ years of age, 1 year after her first surgery. E-G, She is shown at 7, 8, and 9 years of age, after continued reduction of excess skin and residual hemangioma. H and I, Two views of her face show good symmetry of the lids and substantial improvement of all residual facial scars.

Vascular Malformations

Vascular malformations are hamartomas with abnormal collections of capillaries, veins, lymphatics, or combinations of several of these.^{4,38} They are present at birth but not always visible at that time. They can grow slowly over the years, and they do not involute. They may be low flow (most commonly) or high flow. These lesions are discussed in depth in Chapter 11. When the extent or nature of the lesion is in question, preoperative imaging should be ordered. This may involve CT, MRI, MRA, or angiography.

Capillary malformations associated with Sturge-Weber syndrome are worthy of note³⁸ (Fig. 34-24). They present with capillary vascular malformation most typically in the upper two divisions of the trigeminal nerve distribution and are often unilateral but may be asymmetrically bilateral. They are associated with intracranial calcifications and seizure disorders. Early treatment with a laser can significantly decrease the prominence of the surface capillary malformation. Whether this affects later soft tissue hypertrophy is not known. Infants with portwine stains should be followed by an ophthalmologist every 6 months until they are 3 years of age and then every year afterward.⁷

Superficial vascular malformations can be treated with laser therapy, whereas deeper venous malformations may require either sclerosis or surgery (Fig. 34-25). Lymphatic malformations and arteriovenous communications affecting the orbit and eyelid can be treated with surgery, sclerotherapy, and/or embolization.⁷



Fig. 34-24 **A**, This patient had a Sturge-Weber malformation involving the right side of his face. He had seizures associated with this condition and multiple calcifications of the dura of the right frontal lobe and a congenital glaucoma of his right eye. **B**, He is shown at the early stages of his reconstruction, with the noninvolved skin of his left forehead used for his nasal resurfacing. A portion of the forehead flap with the hair-bearing scalp was used for his right brow. His upper lid was resurfaced with a medium-thickness graft from his upper inner arm. His cheek and temporal area were resurfaced with a deltopectoral flap, which was initially attached behind his right ear. The patient did not want to have further reconstruction beyond this stage. In addition, follow-up is no longer possible. (Courtesy of S. Anthony Wolfe, MD.)

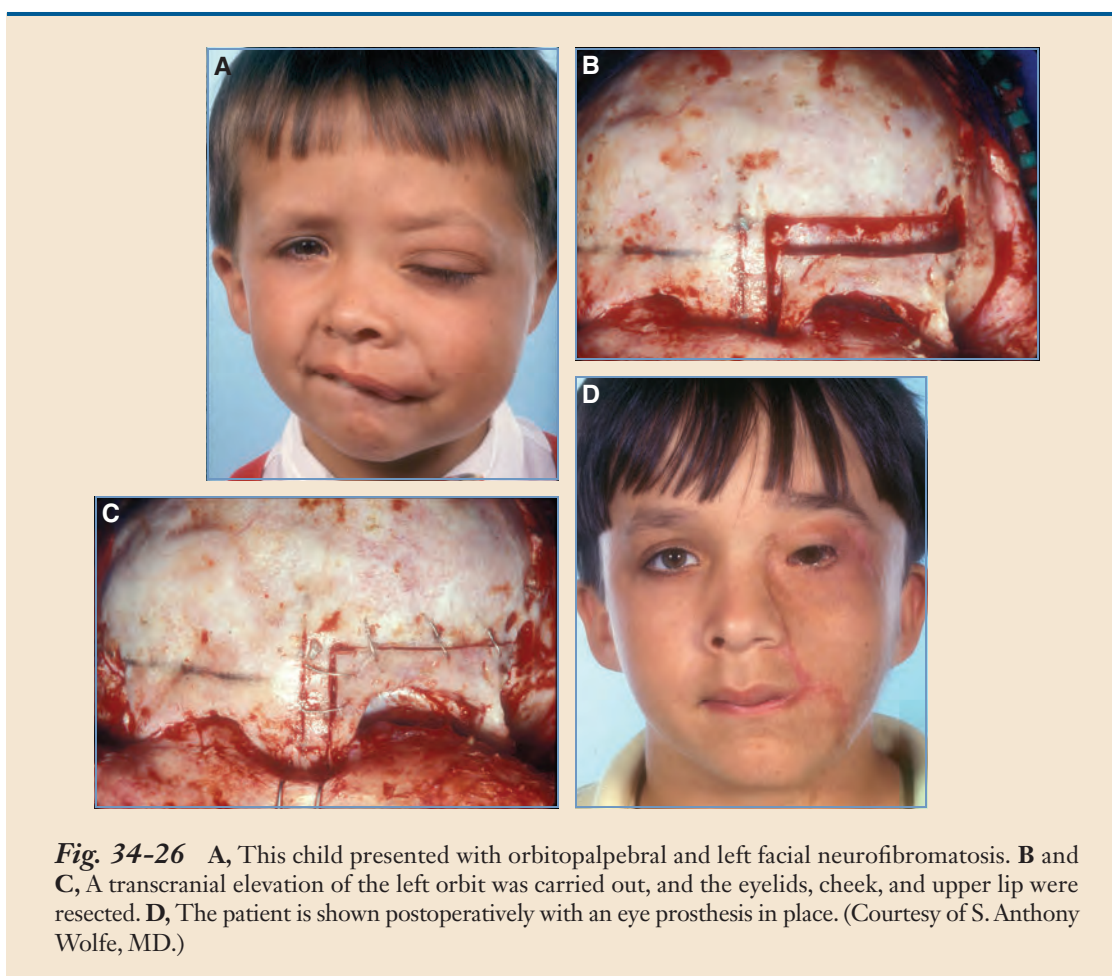


Fig. 34-25 A and B, Frontal and submental views of a young woman with a venous malformation of the left orbit. C and D, Coronal and axial MRIs of the malformation. E and F, The frontal and submental views after the malformation was approached transcranially and the orbital roof removed to provide access to the mass in the superomedial portion of the orbit. The mass was removed in continuity with the orbital floor portion by careful bipolar cauterization of all vessels. (Courtesy of S. Anthony Wolfe, MD.)

Neurofibromatosis

Orbitopalpebral involvement is common in type I neurofibromatosis. A spectrum of deformity exists, ranging from a small, isolated neurofibroma in the eyelid to conditions of such severe deformation that orbital exenteration is the best solution. Optic nerve gliomas, enlargement of the superior orbital fissure, and major defects of the orbital roof with intracranial extension of plexiform neurofibromas may also occur.⁴

Neurofibromas pose a surgical challenge, because they are often vascular and infiltrative with nondistinct borders. Although an en bloc total excision is preferable to debulking, it is rarely possible. The most common form of orbitopalpebral plexiform neurofibromatosis involves an insidious growth with gradual skin changes, loss of skin and supporting tissue elasticity, and ongoing orbital changes. The patient and surgeon should expect that a number of procedures, at times radical, may be required. Rarely is all abnormal tissue removed, and what is not is expected to regrow (though possibly to a lesser extent) (Figs. 34-26 through 34-28).



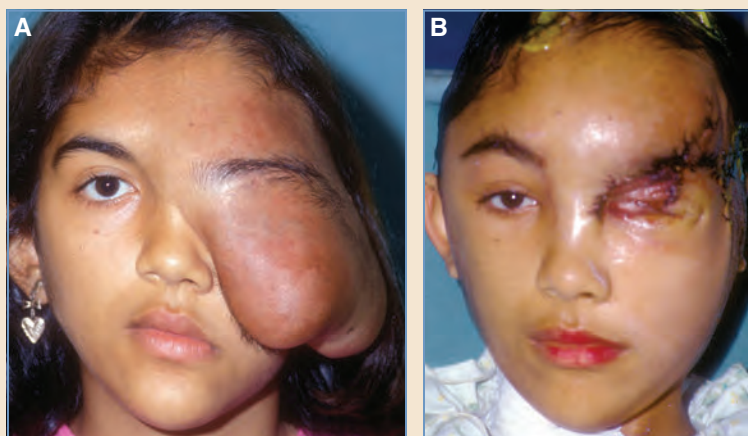


Fig. 34-27 A, This young Cuban child presented to Drs. Paul Tessier and Henry Kawamoto with orbital palpebral neurofibromatosis and was operated on in Havana. B, A transcranial approach was used. A new orbital floor was made out of iliac bone, and the orbital roof was reconstructed with iliac bone. No further follow-up is available. (Courtesy of Paul Tessier, MD.)



Fig. 34-28 A, This young girl presented with orbitopalpebral neurofibromatosis beyond the stage at which reconstruction was thought to be worthwhile. B and C, The eyelid and orbital contents were amputated, and she uses a patch. (Courtesy of S. Anthony Wolfe, MD.)

Congenital Melanocytic Nevi

Congenital melanocytic nevi of the eyelid are uncommon and may present confined to the eyelid alone or as part of a more extensive nevus of the periorbital region, hemiface, or bilateral face. These lesions may be confined to a single lid or present as a synchronous lesion (a so-called divided nevus). The previously described embryology of the eye and eyelids helps to explain the pathogenesis of the divided nevus, because the migration or differentiation of melanocytic cells seems to occur while the eyelids are fused. When the eyelid's open, the nevus appears divided.⁴

The risk of malignant transformation in giant congenital nevi is well established. The risk of malignant transformation in congenital facial nevi appears to be less common; however, the psychological impact on a child with a pigmented facial nevus and his or her family can be profound.

Additionally, eyelid nevi can produce a variety of functional problems. Ptosis may occur as a result of heavy nevus burden on the upper lid. Exophytic growth can produce warty excrescences that cause redundancy in the lower lid with resultant ectropion. Ciliary growth can become disorderly and lead to chronic corneal irritation. For all of these reasons, excision and reconstruction of these nevi seem indicated.

The timing and staging of the excisions vary depending on the extent of the nevus, whether the contiguous nevus needs treatment as well, and the surgeon's comfort in performing the varied flaps and grafts needed. Unlike the excision of malignant tumors of the eyelid, the excision is confined to the skin and subcutaneous tissue; therefore the surgeon must balance the risk of functional problems associated with complete excision, including the full ciliary margin of the lid, with the limited aesthetic deformity associated with a thin residual strip of nevus on an otherwise normal lid margin. If functional issues are involved, the surgeon may decide to resect and reconstruct the full lid margin (Figs. 34-29 and 34-30).

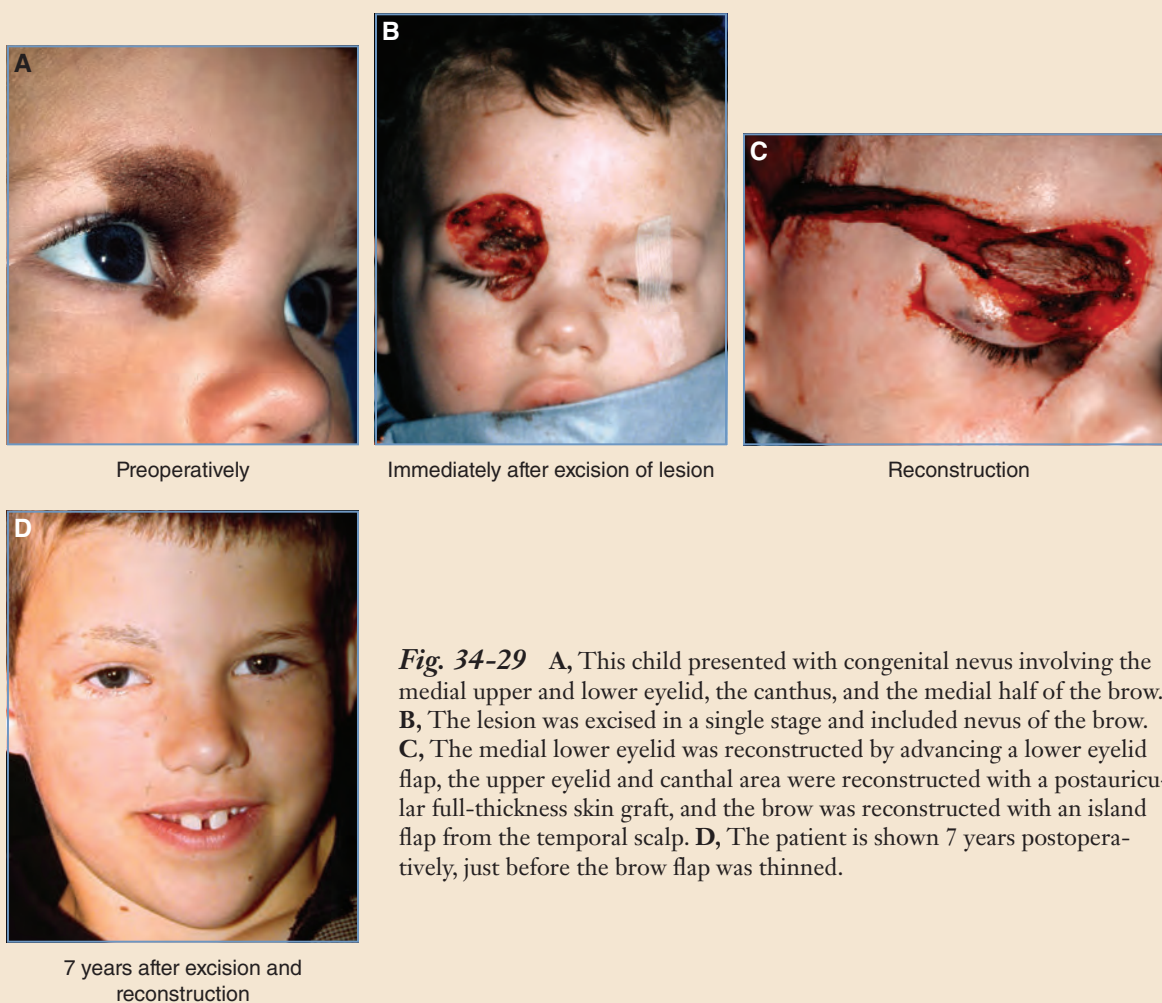
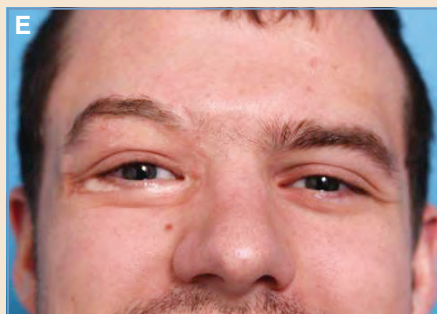


Fig. 34-29 **A**, This child presented with congenital nevus involving the medial upper and lower eyelid, the canthus, and the medial half of the brow. **B**, The lesion was excised in a single stage and included nevus of the brow. **C**, The medial lower eyelid was reconstructed by advancing a lower eyelid flap, the upper eyelid and canthal area were reconstructed with a postauricular full-thickness skin graft, and the brow was reconstructed with an island flap from the temporal scalp. **D**, The patient is shown 7 years postoperatively, just before the brow was thinned.



22 years after excision and reconstruction



3 weeks after brow suspension and scar revision

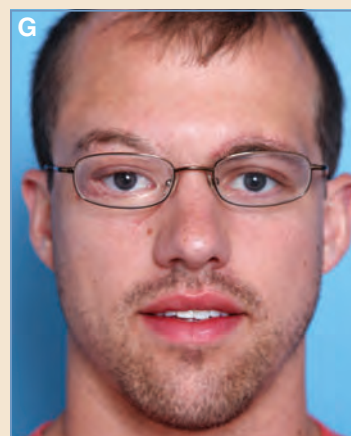


Fig. 34-29, cont'd E, Seventeen years after the thinning of the brow and 22 years after the nevus excision, the graft of the lids is barely perceptible. The brow hair density is quite similar to that of the normal brow, although the normal, medial brow appears ptotic compared with the reconstructed side. F and G, He is shown 3 weeks after elevation and suspension of the left brow and revision of the scar of the lateral eyelid. His lid shape, position, and symmetry are excellent.



Fig. 34-30 This case presents the use of expansion to allow harvest of a full-thickness skin graft from an expanded supraclavicular donor site to cover the eyelids and periorbital area in a single unit. A portion of the expanded forehead flap was used to cover the nasal dorsum after a nevus excision. A, This baby girl presented at 10 months of age with a giant nevus of the forehead, periorbital area, eyelids, and nasal dorsum. Two expanders were placed (250 cc in the forehead and 70 cc in the supraclavicular area). The expanded forehead flap was used to reconstruct the involved forehead and the nasal dorsum. A full-thickness skin graft from the expanded supraclavicular donor site was used to reconstruct the remaining periorbital region. B, The patient is shown after 9 weeks of expansion of the forehead. C, Following a pattern of the lids and the periorbital area, a full-thickness graft was harvested off of the dome of the expander, the capsule was excised, and the donor site was closed. In addition, the nevus was excised, sparing the lid margin and ciliary border and eyebrow. The graft was sutured in place in a single unit. D, The patient is shown 9 years after the excision and reconstruction.

Acquired Deformities of the Eyelid

ACQUIRED LESIONS OF THE EYELID

Relatively few acquired lesions and cysts of the eyelids are seen in children. (Most are discussed in Chapter 8.) Chalazion lesions are one of the few types unique to the eyelids.

Chalazion

A chalazion results from obstruction of the eyelid meibomian glands. Sebum accumulates and instigates an inflammatory response. A chalazion may subsequently become infected.³ Treatment involves the use of warm compresses. Topical corticosteroids and antibiotics may be indicated. If a lesion persists, incision and drainage are indicated. Recurrence is reduced with the injection of corticosteroids afterward.²⁴

POSTTRAUMATIC DEFECTS OF THE EYELIDS

Preoperative Assessment and General Principles

In a trauma patient, visual acuity and ocular motility and the condition of the cornea should be evaluated. For all but the simplest of lacerations, having all patients evaluated by an ophthalmologist is probably prudent. The eyelids require careful evaluation for any foreign bodies and debris. Lacerations involving the eyelid skin with or without the orbicularis muscle are considered superficial. Surgeons should document whether the levator muscle and orbital septum are intact.¹¹

When trauma to the canthus and canalicular system has occurred, the presence of traumatic telecanthus and possible division of the lacrimal duct system is documented. In cases of severe trauma, the globe must be protected with adequate lubrication before and during the reconstruction.

Whether treatment involves limited lacerations of the lid margin or more significant defects, meticulous approximation of the key lid structures is essential to limit secondary defects. Traumatic avulsions of the eyelid from dog bites, human bites, and other accidents require suture approximation and can be successful even 28 hours after the injury. If the tissue loss is severe or tissue has been completely destroyed, there is risk of corneal exposure and subsequent keratinization with vision loss. The risk is greatest when the upper lid is involved, because the upper lid provides the greatest corneal protection.

Eyelid margin laceration repairs are reassessed in 24 hours. The skin sutures are removed after 3 to 4 days. Steri-Strips are applied. Marginal sutures remain in place for 1 week. When the eyelid has been destroyed, repeated observation is necessary to assess for corneal changes. Lubrication is essential during the healing process and until the globe is adequately protected with the varied reconstructive maneuvers.^{9,42-45}

Basic Surgical Principles in Eyelid Reconstruction

Intraoperatively, the eye should be protected by frequently applying lubricants or a balanced saline solution onto the cornea or in the conjunctival cul-de-sac. This is done using a plastic protective corneal lens and retractors or malleables to protect the eye from sharp instruments. Surgeons should select sutures that will prevent corneal irritation. If the eyelid margin is involved, precise alignment is required with appropriate tension. Therefore the use of loupes or other magnifying devices is strongly recommended. Wound edges may be trimmed slightly to freshen them.^{4,9,43-45}

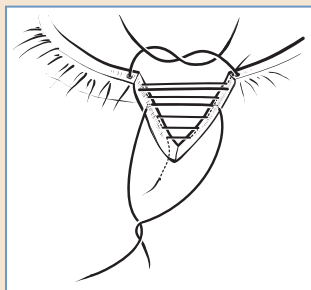


Fig. 34-31 Repair of a full-thickness lower eyelid defect (detail). The conjunctival layer is closed with a rapidly dissolving suture such as 6-0 plain catgut or 7-0 Vicryl. Key sutures are then placed: a 6-0 silk suture at precisely the same level in the gray line, just behind the lash line and a 6-0 Vicryl suture through the tarsal plates.

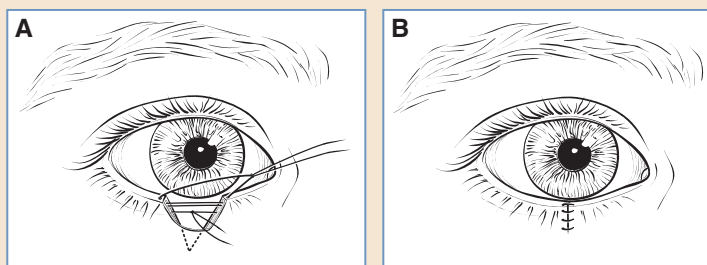


Fig. 34-32 A, A defect involving less than 25% of the eyelid in a child or up to 30% in an adult can be closed by primary repair. B, After the repair.

Upper and Lower Eyelid Defects

Defects of Less Than Twenty-five Percent

A loss of up to 25% of either the upper or lower eyelid can be closed by direct approximation; the figure increases to 30% or more in older patients with some lid laxity (Figs. 34-31 and 34-32). To repair a full-thickness eyelid defect, the tarsal plate should be closed with 6-0 Vicryl as a separate layer, the conjunctival layer with 6-0 plain catgut or 7-0 Vicryl, and the skin with 6-0 silk. The gray line, just posterior to the lashes, should be carefully aligned with 6-0 silk.

Upper Lid Defects of Greater Than Twenty-five Percent

A lateral canthotomy makes it possible to close up to about 40% of the eyelid in combination with the direct closure noted previously (Fig. 34-33). Adding an extension into the temporal area beyond the lateral canthus with a limited rotation flap (Tenzel) (Fig. 34-34) or all the way to the preauricular area (Mustardé) (Fig. 34-35, A) facilitates reconstruction of the outer lamella of the entire lower eyelid. The inner lamella will need to be reconstructed with a rigid mucosa-covered graft, either a split palatal or a septal chondromucosal graft (Fig. 34-35, B and C).

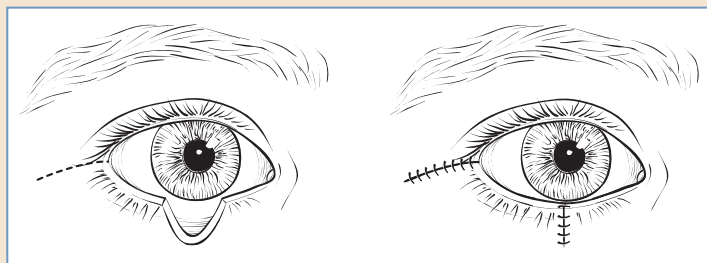


Fig. 34-33 A lateral canthotomy is added if tension on a primary closure seems excessive; this will allow closure of a defect of up to 40% of the lower eyelid.

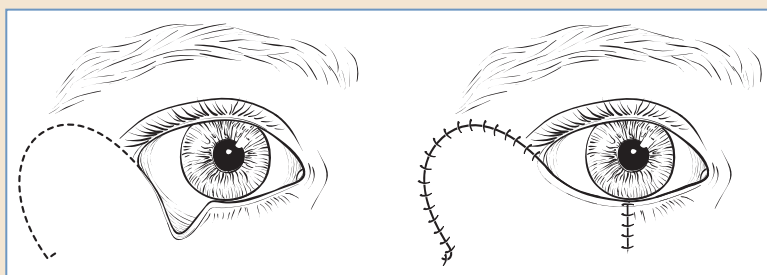


Fig. 34-34 If a lateral canthotomy does not provide enough laxity, the next step is a Tenzel flap, a short rotation flap that does not extend much beyond the lateral rim. Here, a small chondromucosal flap is added for the inner lamella; this can be from the nasal septum, the upper lateral nasal cartilage, or a split palatal graft. (Mucosal grafts alone are not used, because they undergo considerable contraction.)

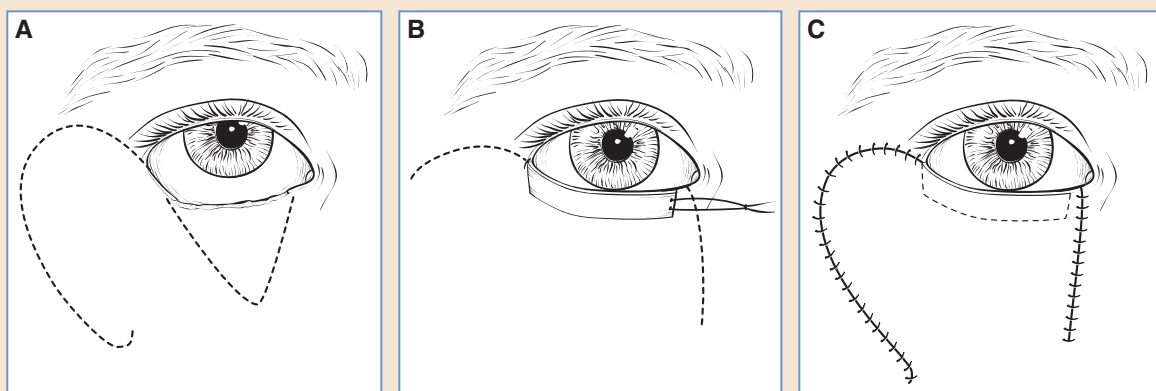


Fig. 34-35 **A**, A full-thickness defect of the entire lower eyelid can be repaired by a Mustardé cheek rotation flap extending to the preauricular area with a cutback. **B** and **C**, This flap relies on a very sturdy septal chondromucosal graft to support the lower eyelid. Its vascularity can be questionable, and it is not our first choice for total lower eyelid reconstruction.

A Mustardé-type cheek rotation flap can be used for total lower lid reconstruction, but we prefer a Tessier nasojugal flap (Fig. 34-36) because of its greater vascularity and support; Mustardé flaps tend to require the rigidity of a thick septal chondromucosal flap to prevent sagging.

Another alternative for defects of most of the lower eyelid is a Kolner procedure (Figs. 34-37 and 34-38). A tarsoconjunctival flap can be taken from the upper lid to reconstruct the defect.

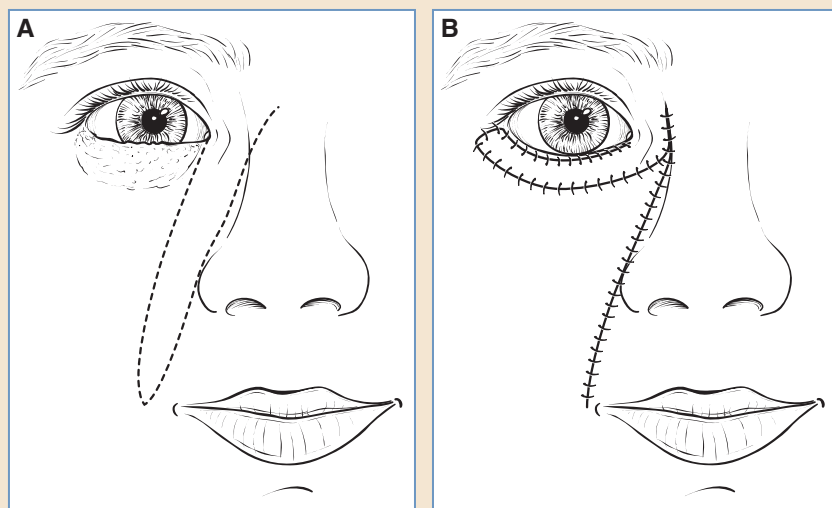


Fig. 34-36 A, Our preferred flap for total lower eyelid reconstruction is a Tessier nasojugal flap. The base of the flap should be slightly higher than the medial canthus, and the tip should be fixed firmly to the periosteum of the lateral orbital rim. B, A chondromucosal or split palatal graft is used for the inner lamella.

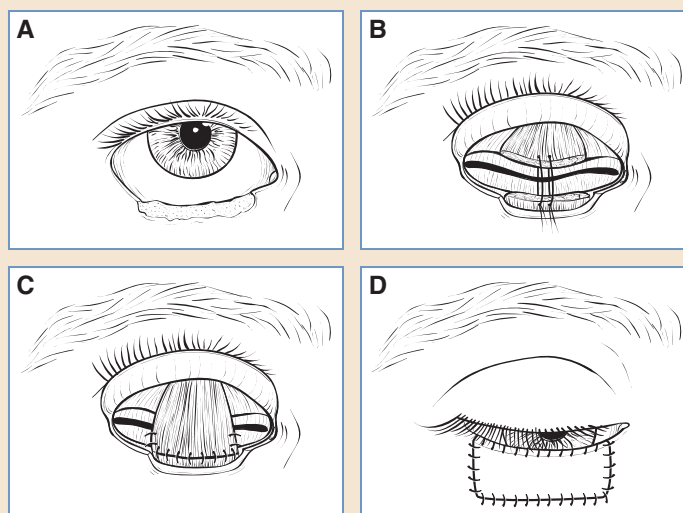


Fig. 34-37 A, For marginal defects of the lower eyelid with a vertical dimension of 10 to 12 mm or less, a tarsoconjunctival flap from the upper eyelid is an excellent alternative. B, The upper eyelid is everted, and an incision is made 4 mm from the lid margin, through the tarsus, to a transverse dimension matching the defect of the lower eyelid. Superiorly, the flap is teased from the overlying levator muscle and contains conjunctiva and Müller's muscle. The dissection should be taken well into the superior cul-de-sac to prevent tension on the upper eyelid when it is sutured into the defect. C, The tarsus of the flap and the remaining native tarsus should be carefully reapproximated. D, A full-thickness skin graft is taken from the preauricular or postauricular area to replace the anterior lamella. (We prefer not to use upper eyelid skin, because it is too thin.) The pedicle of the flap is divided at about 3 weeks, with care to bring the conjunctiva over the lid margin. The donor area will heal spontaneously.



Fig. 34-38 A, This patient presented with an extensive basal cell carcinoma of the lower eyelid and adjacent cheek. B, Resection required removal of the patient's entire lower eyelid and a good portion of his cheek. Reconstruction was divided into two separate segments: the lower eyelid and the cheek. C, A tarsoconjunctival flap was brought from the upper lid and sutured to the remnants of the conjunctiva, carrying with it some tarsus. D, This was covered with a full-thickness graft, and the cheek defect was corrected with the use of a rotation cheek flap. E, The postoperative result. (Courtesy of S. Anthony Wolfe, MD.)

The tarsus is split 3 mm superior to the ciliary margin, the length of the lower eyelid defect. The conjunctiva and Müller's muscle are bluntly dissected from the overlying levator muscle well up into the conjunctival cul-de-sac. The tarsus is sutured to the tarsal remnants of the lower eyelid, and the outer lamella is reconstructed with a full-thickness skin graft.

Upper Lid Defects of Greater Than Fifty Percent

Larger upper eyelid defects require reconstruction, which may involve a selective lateral cantholysis of the lateral canthal superior crus and medial transposition of the upper lid. A lateral cantholysis is recommended to facilitate closure of these wounds, especially those with loss of tissue in the middle portion of either lid. This method releases the tension that would be present with direct closure. Scissors are used to perform a lateral canthotomy. The conjunctiva is separated from the lateral canthal tendon, and a vertical incision is made into the upper or lower crus (depending on the defect). This maneuver may provide 5 to 10 mm of horizontal length. A local skin flap may also be needed. Defects as large as 60% of the horizontal length might necessitate lid-switch procedures. Tissue can be borrowed from the lower lid, whose blood supply is the marginal palpebral artery (Fig. 34-39). These flaps can be 25% narrower than the width of the defect and should be 5 to 6 mm in height to preserve the blood supply. Larger defects (60% to 100% of the length) are often reconstructed using a Cutler-Beard flap or a Mustardé total lid flap. Both are two-stage operations. A Cutler-Beard flap involves creating a full-thickness lower lid flap. The flap is advanced under the margin of the lower lid to correct the upper lid defect. A Mustardé lower lid flap involves a full-thickness lid switch flap and lateral canthotomy. Although the resultant lower lid defect can be corrected with a graft and cheek advancement flap, we prefer to reconstruct the lower eyelid with a Tessier nasojugal flap (Figs. 34-40 and 34-41).

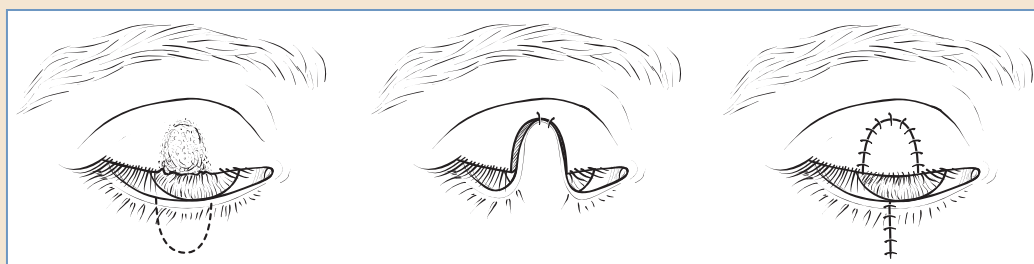


Fig. 34-39 For a partial-thickness defect of the upper lid, either a coloboma or a traumatic defect, the lower lid is the preferred donor area. A Mustardé-Abbé type flap can be based on a very small marginal pedicle; the flap from the lower lid is designed to be a bit smaller than the upper lid defect (a defect of about 20% of the upper lid could be closed primarily). The pedicle is divided at about 3 weeks, with a careful alignment of both lid margins.

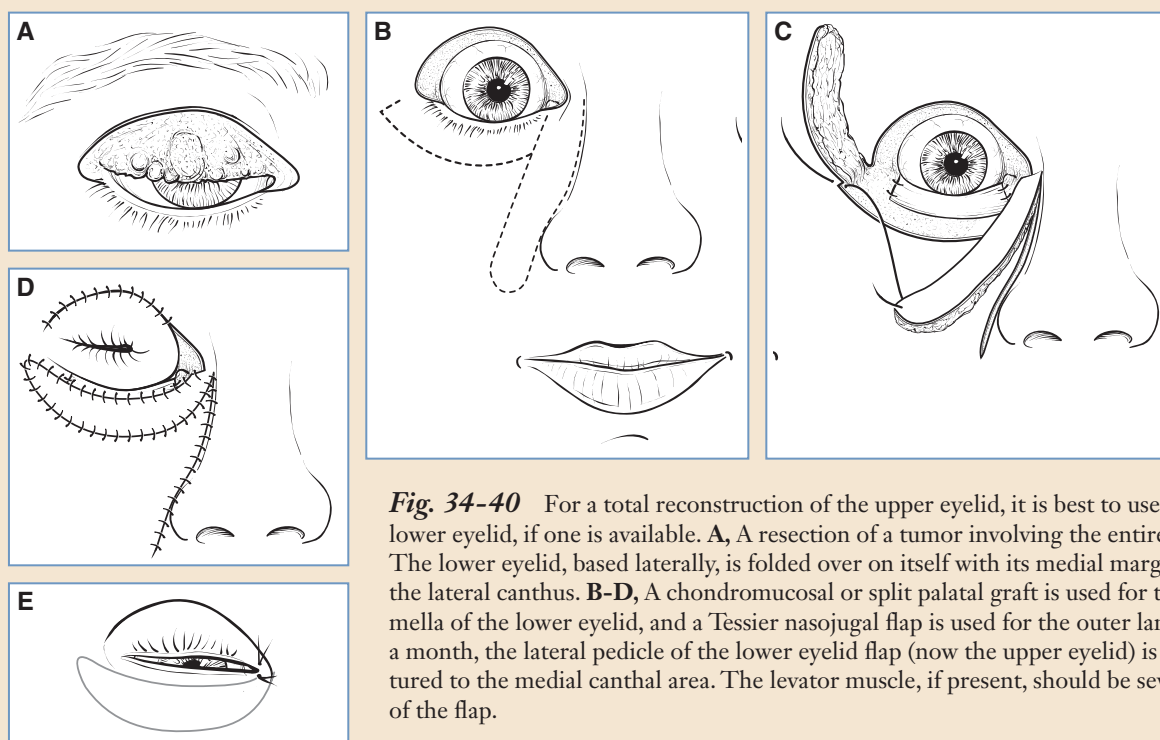


Fig. 34-40 For a total reconstruction of the upper eyelid, it is best to use the entire lower eyelid, if one is available. **A**, A resection of a tumor involving the entire upper eyelid. The lower eyelid, based laterally, is folded over on itself with its medial margin sutured to the lateral canthus. **B-D**, A chondromucosal or split palatal graft is used for the inner lamella of the lower eyelid, and a Tessier nasojugal flap is used for the outer lamella. **E**, After a month, the lateral pedicle of the lower eyelid flap (now the upper eyelid) is divided and sutured to the medial canthal area. The levator muscle, if present, should be sewn to the tarsus of the flap.

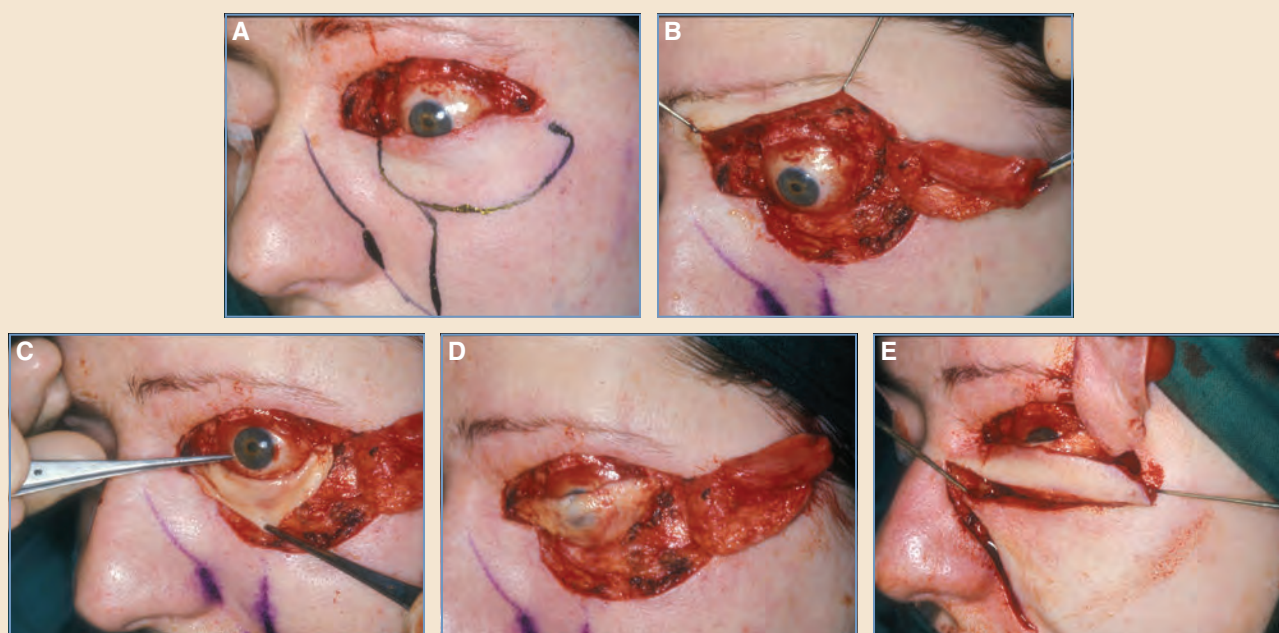


Fig. 34-41 This case describes a total reconstruction of the upper eyelid using the entire lower eyelid, with a nasojugal flap with a buccal graft for reconstruction of the now-absent lower eyelid. **A**, The upper eyelid defect is visualized and the nasojugal and Mustardé flaps outlined. **B**, The lower lid flap is reflected. **C and D**, The buccal graft is placed. **E**, The nasojugal flap is positioned for external coverage of the lower eyelid, and the Mustardé flap is rotated upward into the upper eyelid defect.

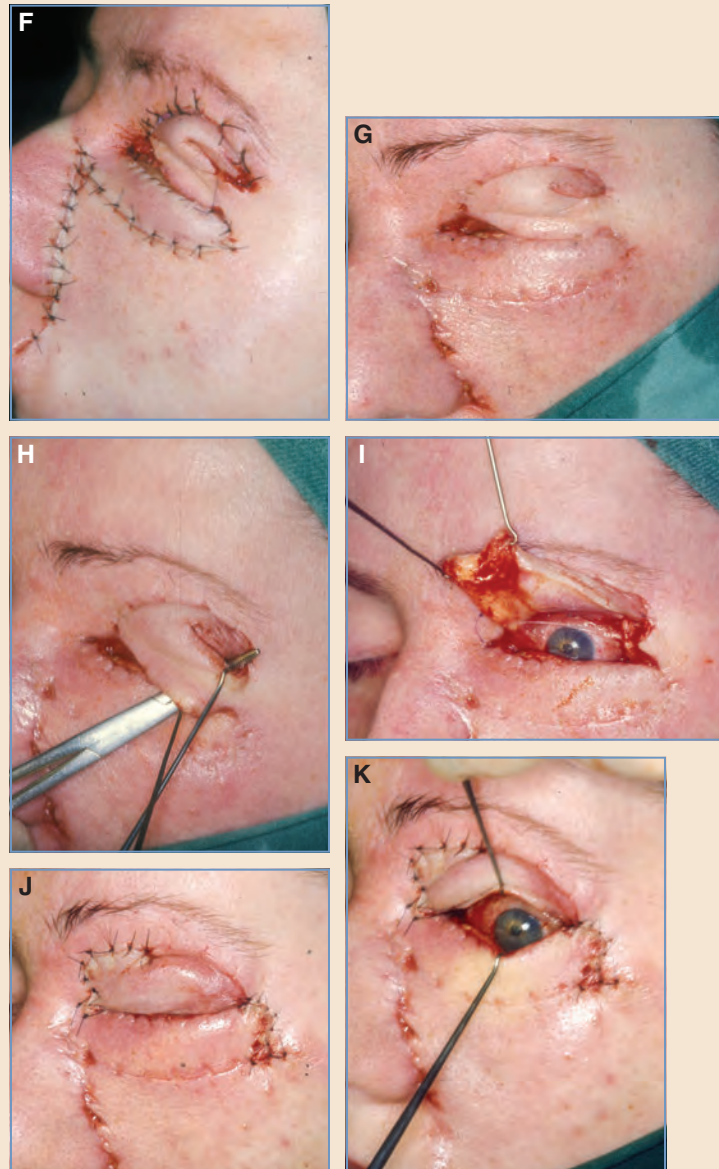


Fig. 34-41, cont'd F, The Mustardé flap is inset, completing the first stage of the reconstruction. G, The flaps are shown 3 weeks after the first stage but before division of the Mustardé flap and inset. H-J, Division and inset of the medial portion of the new upper eyelid. K, Reflection of the completely reconstructed upper eyelid at the completion of the second stage. (Courtesy of Paul Tessier, MD; photos provided by S. Anthony Wolfe, MD.)

Other techniques include a glabellar or nasolabial flap with mucosal or composite grafts. The upper eyelid is essential to protect the cornea. Therefore lower lid reconstruction does not borrow tissue from the upper lid. Tissue may be borrowed from the contralateral lower lid or other areas such as the cheek.^{7,45}

For reconstruction of both eyelids, a very large split palatal graft is sutured to the remnants of the conjunctiva, and the entire defect is covered with a distant flap (posterior forehead, if available), with a small opening medially for drainage. The flap can then be debulked and replaced with medium split (upper lid) and full-thickness skin grafts (lower lid) before division into two eyelids. Movement may be minimal if there is no levator, but if a functioning Bell's phenomenon is present, the eye will be protected and can be of use (Fig. 34-42).



Fig. 34-42 A, This young boy was involved in an automobile accident in South America and had avulsion of both eyelids, much of his nose, and most of his forehead. B, As an emergency procedure, an ear, nose, and throat surgeon placed a pectoralis flap over the entire eye. C and D, The boy is shown after replacement of the pectoralis flap to the chest, a tissue expander to the cheek, a Tagliacozzi flap for the nose (the forehead was not usable), and multiple split-thickness skin grafts of varying thickness for the eyelids. He has minimal levator function but has protective corneal covering and normal vision in his eye. The arm flap is a poor color match for his face but can be resurfaced with a thin split-thickness skin graft, which usually provides a near-perfect color match. (Courtesy of S. Anthony Wolfe, MD.)

Treatment of Secondary Posttraumatic Deformities of the Eyelids

Posttraumatic Telecanthus

There is no better time to correct disruption of the medial canthal tendon than at the initial repair. More extensive trauma to the periorbital tissues and orbit can involve soft tissue and bone. In these cases, obtaining an ideal correction—particularly for unilateral cases—is challenging (Figs. 34-43 through 34-45). The treatment of telecanthus associated with more extensive maxillofacial trauma is discussed in Chapter 40. The actual maneuvers of fixation are similar to those demonstrated earlier in our discussion of the repair of ocular clefts.^{9,42,45} A few additional comments are in order here and pertain to secondary correction.



Fig. 34-43 A, This 3-year-old boy was badly mauled by a pit bull. The medial canthal tendons were avulsed through the laceration in the forehead. B, These were sutured back in position to the periosteum at the primary repair, but this did not heal. C-E, The patient is shown postoperatively after bilateral transnasal medial canthopexies, in which the transnasal wires were tied over a 2 mm miniplate. Lead plates are tied over an Adaptic dressing to compress the skin above the medial canthal tendon. (Courtesy of S. Anthony Wolfe, MD.)

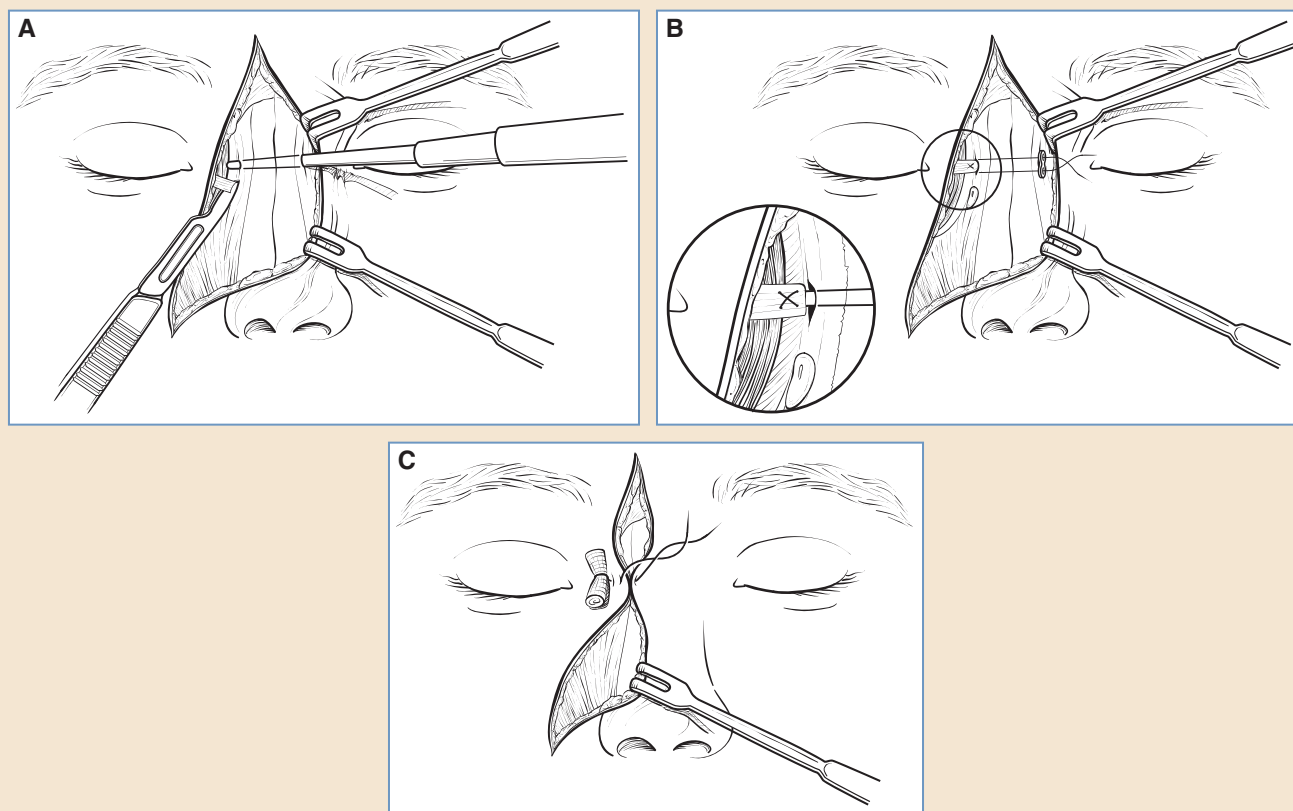


Fig. 34-44 A transnasal medial canthopexy showing the fine details of the passage of the nasal trocar and fixation of the ligament. In the case presented in Fig. 34-43, the procedure was performed for a patient with unilateral nasal agenesis. **A**, The medial canthal tendon is identified and divided from its abnormal attachment. The orbital septal attachments to the upper and lower orbital rims must be extensively divided. **B**, A transnasal hole is prepared with a Tessier canthopexy awl (similar to an ice pick, but with a cutting tip). The *inset* shows the suture. **C**, A canthopexy suture of 2-0 or 3-0 stainless steel wire is then passed through the hole (retrieved with a loop from the contralateral side) and tied over a two-hole titanium miniplate as a toggle.

Excellent results are virtually never obtained, particularly if they are compared with a normal canthus on the contralateral side. A good result requires extensive subperiosteal dissection of the orbital septal attachments and careful identification of the medial canthal tendon through a skin incision made close to the tendon. A wire suture is passed twice through the tendon, passed through a transnasal hole, and tightened over a two-hole miniplate on the contralateral side. Additional transnasal wires above the medial canthopexy can be passed by placing a spinal needle on a K-wire driver and then passing a transnasal wire; this is done twice, and the wires are tightened over Adaptic pledgets (see Figs. 34-43 through 34-45). This helps to prevent recurrent thickening and deepens the paranasal hollow anterior to the medial canthal attachment, which is important in obtaining a good result.

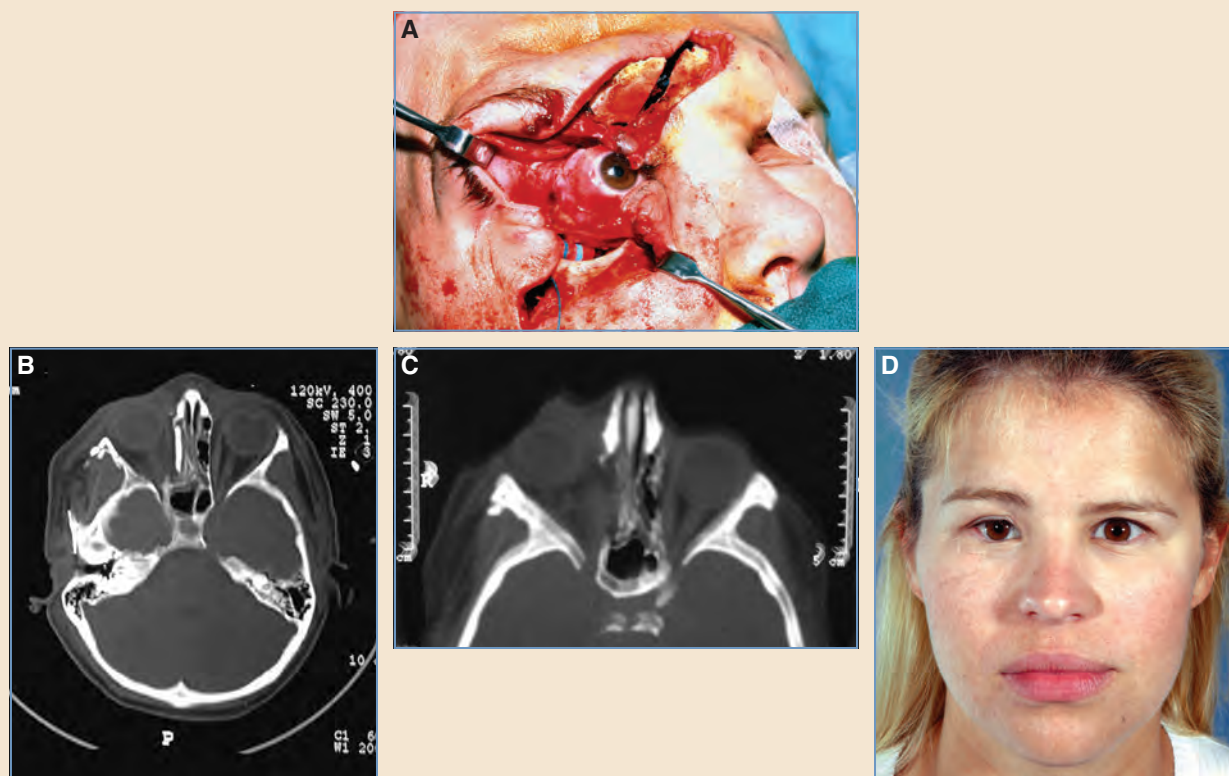


Fig. 34-45 This young girl was run over by a motorboat propeller. **A**, Her upper and lower eyelids were lacerated and partially avulsed, and the propeller passed through the anterior cranial base. The globe was preserved. **B** and **C**, Two axial CT views show the associated orbital wall injuries. **D**, The patient is shown postoperatively after reduction and fixation of the zygomatic fracture; cranial bone grafts to the orbital roof, medial wall, and floor; and meticulous repair of the eyelid injuries. The medial rectus muscle was mildly damaged. The patient may require an adjustment of the medial and lateral rectus muscle insertions. (Courtesy of S. Anthony Wolfe, MD.)

Correction of Secondary Lower Eyelid Laxity and Retraction

Lower eyelid retraction can be caused either by laxity—or retraction with an adequate amount of skin—or a skin deficiency. In both instances correction can be difficult, and on occasion it can require several stages.^{9,43-45}

With minimal laxity and adequate skin, a lateral canthopexy may be adequate. This can be performed through a coronal incision (if one is used for other reasons) or through an upper eyelid incision (lateral retinacular canthopexy). A more powerful canthopexy can be performed by developing a dermoorbicular pennant flap, bringing it through a burr hole in the lateral orbital rim, and sewing it to the temporal fascia (Fig. 34-46; see Fig. 34-10). When skin is deficient, a laterally based flap of upper lid skin or a full-thickness graft can be used.

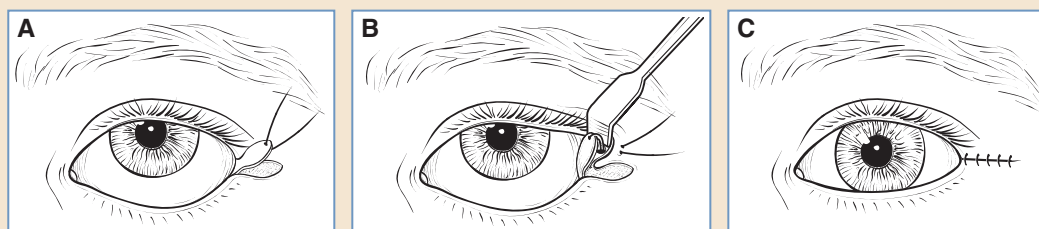


Fig. 34-46 The steps in a lateral canthopexy. Tarsal strip techniques are generally not useful in children because of insufficient lid laxity. The dermoorbicular pennant procedure, however, can provide a very strong lateral canthopexy. **A**, The flap is extended laterally from its base on the upper and lower eyelids and deepithelialized. **B** and **C**, A 3 to 4 mm hole is drilled through the lateral orbital rim in the desired position of the canthus. The flap is passed through this hole and sutured to the periosteum of the lateral portion of the orbital rim.

If the soft tissue of the lateral canthal area is of poor quality (as might be the case in a patient with neurofibromatosis or blepharophimosis), a palmaris longus or short toe extensor tendon graft can be taken and split into a Y, with two limbs sutured to the tarsal plates. After 5 to 6 months, it can be used in a similar fashion to the dermoorbicular pennant.

Mild eyelid retraction with good canthal position and adequate skin can be treated successfully with a split palatal graft.

CONCLUSION

The management of malformations, neoplasms, and trauma of the eyelids in children differs little from that in adults, with the proviso that any condition preventing light and images from reaching the retina must be addressed immediately to prevent the development of amblyopia. Lid laxity and excess skin are rare in children; these are variables that could be used for correcting many defects.

Colobomas of the upper eyelid that result in a lack of corneal protection need to be corrected immediately to prevent corneal exposure and scarring. However, in congenital facial palsies such as Moebius syndrome, which involves lagophthalmos, this seems to be much better tolerated by children than adults.

The principles of eyelid reconstruction have been outlined: the inner lamella must be reconstructed with a structure that is both rigid and covered with mucosa. Only septal and upper lateral cartilage chondromucosal grafts and split palatal grafts fulfill this requirement. Outer lamella or skin can be provided from a number of sources. We find that the medium-thickness skin grafts from the upper inner arm or skin from the contralateral upper eyelid is best for the upper eyelid; the lower eyelid, which has no mobility requirements, can be covered with full-thickness skin grafts from the retroauricular or other regions. When possible, the levator should be attached in upper eyelid reconstructions.

Finally, close collaboration with a pediatric ophthalmologist is important in most cases.

KEY POINTS

- Congenital and acquired deformities of the eyelids in pediatric patients present a unique and complex treatment challenge for plastic surgeons.
- Corneal protection is paramount. Several days of treatment with corneal lubricants and a wet chamber is possible, but a cornea that remains exposed and shows signs of desiccation is a surgical emergency.
- The risk of amblyopia is a major consideration in pediatric patients with eyelid deformities. Conditions that either obstruct the visual axis (deprivation amblyopia) or distort the globe (anisometropic amblyopia) necessitate urgent care. Patients in whom amblyopia is suspected need to be referred to an ophthalmologist for evaluation.
- An interdisciplinary approach (plastic surgeon, ophthalmologist, and neurosurgeon) may be necessary for complex eyelid or craniofacial malformations.
- Eyelid reconstruction of any sizable defect must at a minimum provide an outer and inner lamellar skin cover, a mucosal structure to be in contact with the cornea, and structural support to mimic a tarsal plate (either septal, upper lateral, or ear cartilage or the inherent rigidity of a split palatal graft).
- The lower eyelid does not require mobility, but the upper eyelid does; therefore the lower lid may be used for upper lid reconstruction (Mustardé cross-lid switch flap), but the upper lid should not be used for the lower lid. (We agree with this as far as using the entire upper lid for the lower lid, but we do find it useful to use tarsoconjunctival flaps from the upper lid for marginal defects of the lower lid.)
- Tarsal strip techniques are generally not useful for lateral canthal tightening in children, because lid laxity is insufficient.
- An algorithm for the treatment of eyelid defects will provide a logical progression of treatment modalities, depending on the size of the defect.

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35

Congenital and Acquired Deformities of the Nose

Fernando D. Burstein • Bruce S. Bauer



As the central and prominent feature of the face, the nose commands front billing, and even minor nasal abnormalities and discrepancies draw immediate attention. There has always been an inherent desire in people to look like their fellow humans and not appear peculiar or different.¹ The complex sequence of events that shapes the frontonasal structures and functional airway is vulnerable to atypical cellular processes and deformational forces that may cause minor abnormalities in nasal shape at one end of the spectrum, or near-total absence of nasal formation at the other end. After the normal nose has formed, its prominent position makes it subject to the forces of trauma, particularly in healthy, active children. Regardless of whether the deformities result from abnormal developmental processes or an errant bat during Little League, there is a need either to create a more normal appearance or restore the structure so that pediatric patients can interact comfortably and successfully with their peers. Although other chapters address nasal deformity resulting from cleft lip and palate (CL/P), the range of nasal clefts in atypical craniofacial clefts (see Chapter 26), and primary rhinoplasty for adolescents with indications and techniques (see Chapter 36), this chapter will review the diagnosis and treatment of some of the more common conditions that may affect the development and/or appearance of the nasal structures. These conditions include nasal hypoplasia, atypical nasal clefts, congenital nasal masses, vascular lesions, cutaneous lesions, and traumatic nasal deformities.

NOSE EMBRYOLOGY

During the fourth week of gestation, neural crest cells migrate to the developing face from the lower forebrain, the midbrain, and rhombomeres 1 and 2 of the upper hindbrain. These migratory neural crest cells are the predominant source of facial connective tissue, including cartilage, bone, and ligaments. Because neural crest cells migrate to the face as cohorts of cells from different portions of the brain, they carry with them different developmental programs. Mutations arising in the premigratory or early migratory neural crest cells may affect a specific clone of cells, which then carries that mutation to a predestined site in the face.²⁻⁶

At week 4, five identifiable primordia surround the stomodeum (Fig. 35-1). The single, unpaired frontonasal prominence lies in the midline just superior to the stomodeum.² By the end of the fourth week, even before the neural folds close, paired thickenings of ectoderm appear on the surface of the frontonasal prominence just superolateral to the stomodeum. These oval nasal placodes, located at the 1 o'clock and 11 o'clock positions, give rise to the future nose and nasal cavities. Development of the nasal placodes (and the lens placodes) requires the presence of the paired box gene *PAX6*. In the absence of *PAX6*, neither the nasal placodes nor the lens placode develops.² During the fifth week of gestation, mesenchyme in the margins of the nasal placodes proliferates to form horseshoe-shaped elevations—the nasomedial and nasolateral processes. The nasomedial processes are longer than the nasolateral processes.³ The tissue surrounding the placodes thickens and elevates so the nasal placodes appear recessed within depressions in the surrounding tissue, which are designated the *nasal pits*. The *nasal pits* are the primordia of the anterior nares (the future nostrils) and the nasal cavities.³

From the fifth week of gestation, the nasal pits gradually deepen toward the oral cavity, forming substantial depressions. By 6½ weeks' gestation, only a thin *oronasal membrane* separates the oral cavity from the nasal cavities. This oronasal membrane then breaks down so that the oral cavity can communicate with the nasal cavity through openings posterior to the primary palate.² These openings are designated the *nasal choanae*. Fusion of the two palatal shelves lengthens the nasal cavity and carries the communication posteriorly to the upper pharynx. The nasal septum grows down from the frontonasal prominence to the level of the palatal shelves when the shelves fuse to form the definitive secondary palate. The fusion of the primary palate with the secondary palate is marked by the incisive foramen.⁷

From the sixth to eighth weeks of gestation, the cheeks and corners of the mouth form by the merging of the maxillary and mandibular processes. The upper lip is completed in the latter part of this period.² Concomitantly, the expanding nasomedial processes merge with the superficial region of the maxillary processes on both sides along epithelial seams called the *nasal fins*.² *Mesenchyme* penetrates the nasal fins and forms continuity between the nasomedial and maxillary processes. The fusion of the two nasomedial processes displaces the frontonasal prominence posteriorly. Therefore the frontonasal prominence does not contribute significantly to the definitive upper lip, jaw, or nasal tip, despite the fact that it formed a prominent portion of the stomodeal border earlier. The fusion of the paired nasomedial processes forms the nasal tip, the crest of the nose, and a portion of the nasal septum.²

Facial and limb malformations are known to result from deficient or excess molecular signaling (such as Sonic Hedgehog, fibroblast growth factor, and retinoic acid signaling). Similar phenotypes, such as clefting, may result from either the deficiency of the appropriate midline tissue or the excess of other midline tissue that the appropriate processes cannot meet to fuse. In experimental animals, reduced retinoic acid signaling diminishes the expression of Sonic Hedgehog and fibroblast growth factor 2 in the mesenchyme, increases apoptosis locally, and decreases prolifera-

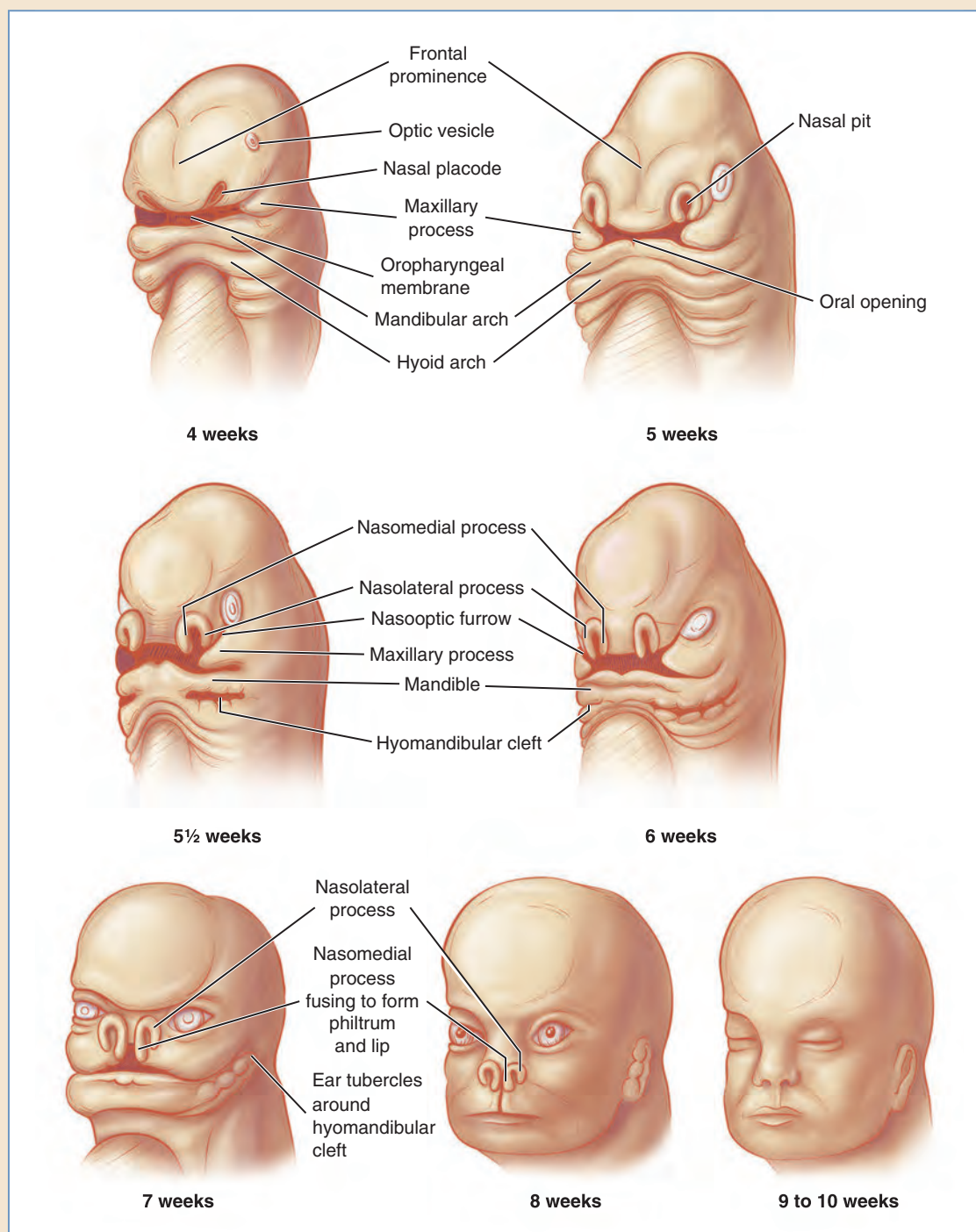


Fig. 35-1 Embryogenesis of the face from 4 to 10 weeks' gestation shows the contribution of the frontonasal process and the developing nasomedial and nasolateral processes to the formation of the nose, while the structure of the lip forms around the developing stomodeum and the nasal pit deepens.

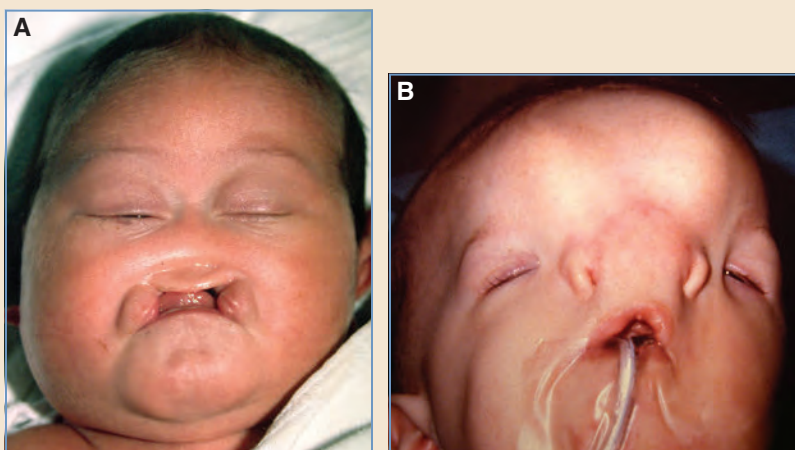


Fig. 35-2 A, Newborn infant with a hypoplastic forebrain, nasal hypoplasia, and absence of the premaxilla, derived from the frontonasal process. Experimentally reduced retinoic acid signaling diminishes the expression of Sonic Hedgehog and fibroblast growth factor 2 in the mesenchyme, increases apoptosis locally, and decreases proliferation of tissue in the forebrain and frontonasal process. Animals with reduced retinoic acid signaling show a holoprosencephalic phenotype similar to what is seen in this neonate. B, Conversely, as shown in this infant with frontonasal dysplasia, excess Sonic Hedgehog stimulates frontonasal growth and widens the frontonasal process, resulting in cleft palate. More severe phenotypes include infants with frontonasal dysplasia or median facial cleft.

tion of tissue in the forebrain and frontonasal process. These animals show holoprosencephalic phenotypes with hypoplastic forebrains, fused eyes, and the absence of structures derived from the frontonasal process.^{2-4,7,8} Conversely, excess Sonic Hedgehog stimulates frontonasal growth and widens the frontonasal process by an average of 48%, resulting in cleft palate, and more severe phenotypes show ectopic midfacial structures with duplication of the nasal bones²⁻⁹ (Fig. 35-2).

FACIAL SKELETON AND NASAL STRUCTURES

The cartilage of the nasal capsule is the foundation of the upper part of the face.¹⁰ The bony elements of the facial skeleton appear around the nasal capsule and partly replace it. The lateral masses of the ethmoid form by endochondral ossification of the nasal capsule. The frontal processes of the maxillary bones, premaxillary bone, nasal bones, lacrimal bones, and palatine bones all form in membrane in close relationship with the roof and lateral walls of the cartilaginous nasal capsule.¹⁰ The vomer develops in membrane in relation to the perichondrium of the septal process. Eventually, nearly all of the nasal capsule becomes ossified or atrophied. All that remains of the nasal capsule cartilage in adults is the anterior part of the nasal septum and the alar cartilages that surround the nostrils.¹⁰

Specifically, the midline septal cartilage is continuous with the cartilaginous skull base. At birth, the skull base has three major ossification centers, and the septal cartilage has not yet ossified. The lateral masses of the ethmoid have ossified, forming the paramedian bones, but the cribriform plate is still cartilaginous or fibrous. Therefore at birth, the entire midline of the face

may be a lucent strip of cartilage situated between the paired lateral ossification centers. This lucent midline can simulate a midline cleft from the nares to the presphenoid bone.¹⁰ Because the appearance of the nasal septum varies with the patient's age, one must carefully interpret imaging "evidence" of midline defects and sinus tracts. In 14% of patients younger than 1 year of age, there is no midline ossification of the anterior fossa or septum.¹⁰⁻¹³

OVERVIEW OF CONGENITAL NASAL ANOMALIES

Losee et al¹³ presented a classification scheme for congenital nasal anomalies based on a review of 261 patients seen over a 22-year period at the craniofacial center at The Children's Hospital of Philadelphia. They separated anomalies into four types:

- Type I: Hypoplasia and atrophy
- Type II: Hyperplasia and duplications
- Type III: Clefts
- Type IV: Neoplasms and vascular anomalies

These types account for 62%, 1%, 16%, and 20% of cases, respectively. The high association of both type I and type III anomalies with craniofacial syndromes may somewhat skew the prevalence of these groups of anomalies in this chapter. Type I anomalies include paucity, atrophy, or underdevelopment of skin, subcutaneous tissue, muscle, cartilage, and bone in varying degrees. Although arhinia and unilateral nostril agenesis are quite rare, lesser forms of nasal hypoplasia are very common as part of many craniofacial syndromes. Type II anomalies involve an excess of tissue and include the spectrum of deformities from bifid septum and columella associated with dermoid cysts and sinuses, to the rare proboscis lateralis. Type III anomalies include Tessier nasal clefts¹⁴ (Tessier 0, 1, 2, and 3) and their cranial extensions (Tessier 11, 12, 13, and 14). These clefts usually result in hard and soft tissue deficiencies and are also frequently associated with other craniofacial syndromes. Type IV anomalies include congenital masses, such as the spectrum of dermoid neoplasms, glioma, encephalocele, vascular malformations, and benign and malignant neoplasms. Although we have taken a slightly different path with the basic sections of this chapter, it may be helpful to keep the Losee types¹³ in mind when one is considering the spectrum of congenital nasal lesions.

Congenital Absence of the Nose

From a review of the previous relatively brief discussion of the very complex processes that occur in the development of the nose, it appears that much rests on the effects of molecular signaling and tissue patterning in the face. Sonic Hedgehog may be the morphogenic organizer, whereas fibroblast growth factors may serve as the stimuli for mesenchymal outgrowth. Insufficiency of the frontonasal and nasomedial processes may result in hypoplasia or absence of the nose and intermaxillary segment (see Fig. 35-2, *A*). If there is decreased proliferation in the forebrain and frontonasal processes, the plumping of the nasomedial and nasolateral processes do not progress or progress incompletely. The ectodermal sculpting that should progress to create the nares, and the gradual breakdown to communicate with the pharynx, may either not progress at all or progress incompletely (Fig. 35-3). Lesser distortions of this process may result in isolated nasal deformities, with incomplete canalization of the nares and residual soft tissue and cartilage masses within the nasal airway. Not surprisingly, given the intimate relationship between the developing brain and central facial structures, the most severe forms of nasal hypoplasia may be associated with brain anomalies (often in the holoprosencephaly spectrum)^{15,16} (see Fig. 35-2, *B*).

Given the fact that newborn infants are obligate nose breathers, children born with either incomplete formation of the nose or total absence of the nose have the same emergent problem as children born with complete choanal atresia, and after immediate intubation, these infants need a tracheostomy. Subsequent reconstruction of an absent nose is complicated by hypoplasia of the supporting tissues (both nasal and maxillary, cartilaginous and bony), soft tissue deficiencies, and the desire to create a functional nasal airway and maintain its patency, if possible. These procedures may need to be staged over many years and are not typically completed until the affected child reaches skeletal maturity (Fig. 35-4). A combination of forehead flaps, nasolabial flaps, and local tissue advancements is necessary for soft tissue coverage.^{17,18} Cranial bone grafts, rib grafts, and conchal grafts can be used for skeletal reconstruction. Microsurgical tissue transfer may be essential in the most severe cases to provide enough tissue for internal lining and external covering.



Fig. 35-3 This infant has a rare combination of nostril agenesis and a complete bilateral cleft lip. The border of the nostril is evident on the left side, but the nasal pit failed to deepen and break through the buccopharyngeal membrane. Consequently, there is a solid mass of tissue in the airway to the level of the piriform margin, where the posterior airway is patent.



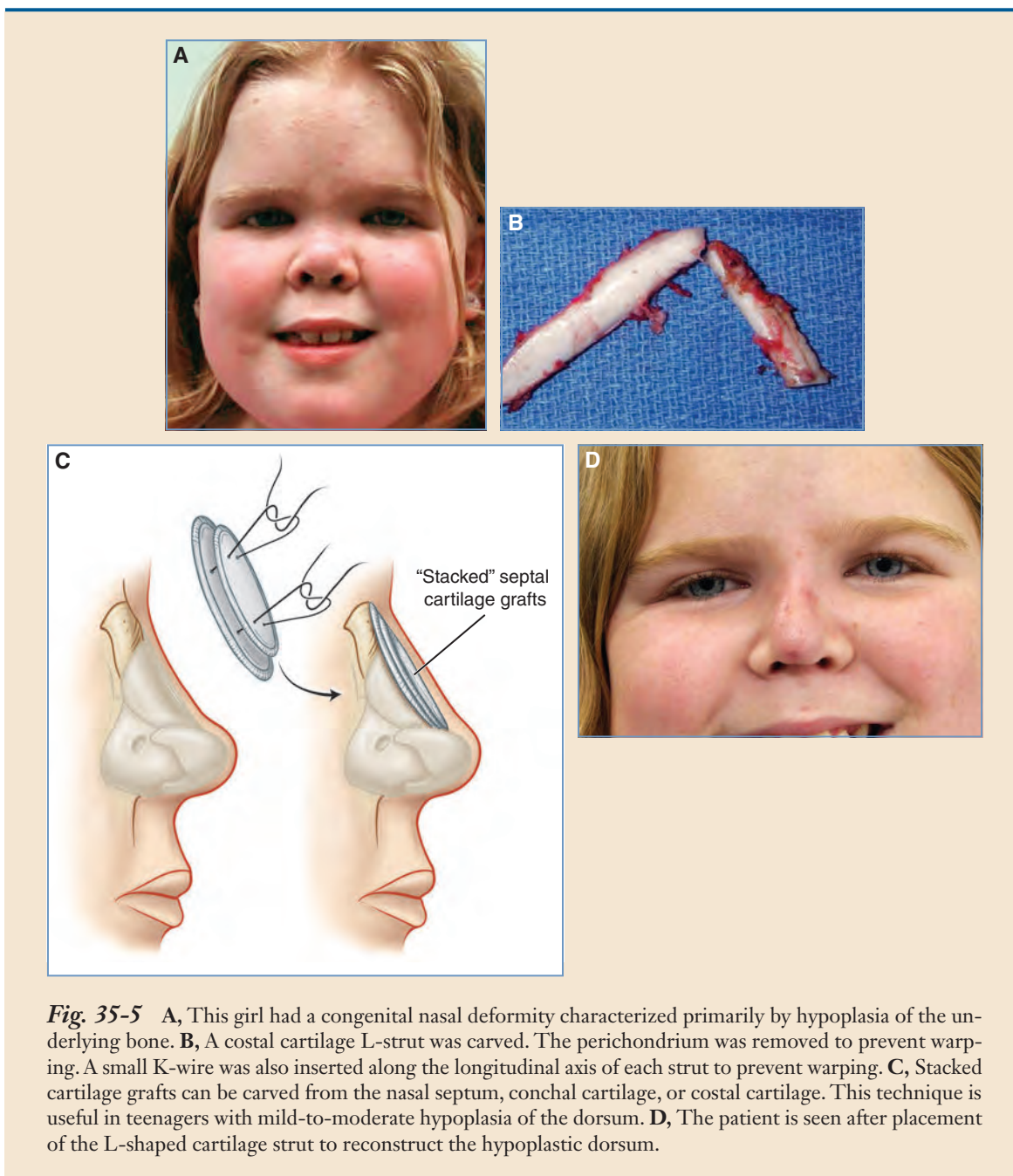
Fig. 35-4 **A**, This child was born with severe nasal hypoplasia, absence of the nasal airway on the right side, a hypoplastic nasal passage on the left side, mild hypotelorism, and a midline cleft of the lip. Additional anomalies were noted in the heart and gastrointestinal tract. She showed no CNS anomalies. During her first week of life, she underwent a tracheostomy. **B**, A three-dimensional CT scan showing frontonasal hypoplasia, medialization of the orbits, absence of a nasal passage on the right side, and presence of a nasal passage at the level of the piriform margin larger than what is seen at the skin level. **C**, After an initial failed attempt to reconstruct the external nose with expanded regional flaps when this patient was 5 years of age, formal reconstruction with expansion of the forehead was begun when she was 9 years of age.



Fig. 35-4, cont'd D, Two weeks before forehead flap reconstruction, regional flaps were elevated around the nostril pit on the left side, and bone was drilled out to open the airway to the point where the local surface flaps could be sutured to lining tissue behind the piriform aperture. E, Tissue expander in place and flaps healed around the reconstructed left nasal passage. F-H, Incisions were made at the borders of the planned external nasal reconstruction, and the border flaps were turned toward one another to provide lining tissue for the left nasal vault and coverage for the combined cranial bone and cartilage grafts used for support beneath the forehead flap. A pseudonostril was created on the right side. Results are seen at I, 14 years of age, and J and K, 19 years of age.

Nasal Hypoplasia

Nasal hypoplasia can present in a number of ways and with varying degrees of deformity. The severity of the deformity may be a function of how early in development the normal processes are interrupted and whether the tissues required to form the individual nasal parts are not developed, partially developed, or present but displaced. Nasal hypoplasia can be associated with dwarfism and other genetic conditions¹¹ (Fig. 35-5; see also Fig. 33-3). Trauma at an early age can also re-



sult in lack of nasal growth. The nasal dorsum may be very low because of a lack of growth of the nasal bones and septum.^{8,9} Less frequently, the tip cartilages themselves may be hypoplastic, with resultant aesthetic and functional sequelae. The severity of the condition may not become fully apparent until skeletal maturation.

A functional airway must be established early, and it is generally advisable to allow time for growth of the nasal structures. Whenever possible in the initial corrective efforts, structures should be placed as aesthetically as possible before the child reaches school age. Typically, bone grafts that are placed early will need to be augmented in later years. We have found that there was growth of these grafts, but the growth was not sufficient to obviate the need for later augmentation. Early grafts may still have the benefit of stretching the overlying skin envelope (see Fig. 35-5). Definitive reconstruction may be delayed until the child is close to skeletal maturity. Reconstruction through an external rhinoplasty approach can yield gratifying results. Autologous cartilage grafts from the septum, conchae, and rib serve as the framework for nasal reconstruction. Wide undermining of the nasal skin to the frontal bone superiorly and the maxilla laterally usually yields enough soft tissue to cover the framework.

Facial Clefts

Deranged development of the frontonasal process and/or failure of adjacent processes to merge successfully results in a coherent series of malformations.^{15,16} The insufficiencies of the previously described frontonasal and nasomedial processes result in some of the common manifestations of holoprosencephaly.^{8,9,11} Failure of the two nasomedial processes to merge in the midline produces the more rare, true midline CL/P with hypertelorism. These defects and the full spectrum of typical and atypical clefts have varied effects on the overall structure of the nose (Fig. 35-6; see Figs. 35-7 through 35-10). A discussion of the typical cleft lip nasal deformity and a review of the classification and features of atypical nasal clefts appear elsewhere in this book. Several different anatomic classifications for rare craniofacial clefts have been reported. In 1976 Tessier¹⁴ proposed a new system of classifying facial and craniofacial clefts based on the orbit as a reference structure. He recognized that there was both a soft tissue and a bony component to these clefts. His system attempts to offer common terminology for the description and treatment of these rare and complex clefts. As noted earlier and described in detail in Chapter 26, the Tessier nasal clefts include clefts 0, 1, 2, and 3, with their cranial extensions being Tessier clefts 11, 12, 13, and 14. Kawamoto¹⁹



Fig. 35-6 This infant was born with bilateral Tessier number 1 through 13 clefts and asymmetrical number 3 clefts without clefting of the lip, associated with a small encephalocele.



Fig. 35-7 A, This 5-year-old boy was born with a Tessier number 2 cleft on the left that was not associated with any bony anomaly. B-E, Outline of the nasal flaps, including both skin and cartilage, used for the cleft repair. F and G, Result 2½ years after the first surgery and 6 months after a revision procedure to modify a limited alar web.

reported that the incidence of these atypical clefts ranged from 1.43 to 4.85 per 100,000 births, making them far rarer than typical CL/P malformations. David et al²⁰ studied these rare clefts with CT and three-dimensional reconstructions, improving our comprehension of the anatomic anomalies and reconstructive challenges.

As with the design and planning of a typical cleft lip repair, the repair of nasal clefts in almost all patients requires rearrangement of the cartilaginous structures (with or without additional cartilage grafts and one or more types of Z-plasty lengthening procedures) to correct the vertical deficiencies inherent in all of these cases²¹ (Figs. 35-7 and 35-8; see also Fig. 35-6). For more complex clefts, repair often requires simultaneous correction of one or both eyelids and the lip (see Fig. 35-9), although other clefts may be confined entirely to the nose. Not surprisingly, most facial clefts need staged revisions as the child grows.^{22,23} Often the defect in these patients has a bony component that involves the maxilla and skull. Reconstructive planning in these complex cases requires bony and soft tissue reconstruction, which is staged as the patient develops. Complex reconstructive techniques, including soft tissue expansion, bone grafting, reconstruction of



Fig. 35-8 A, This child with Fraser syndrome, a rare autosomal recessive disorder, shows the features of cryptophthalmos and nasal anomalies, including a broad nose with a midline groove, depressed nasal bridge, and hypoplastic nares with Tessier number 3 clefts. B and C, Outline of Z-plasty to reposition parts of the disrupted alar margins carried out when the child was 4 years of age. D, Appearance 2 years after repair. E and F, Appearance at 12 years of age.

the lacrimal system, and on occasion craniosynostosis surgery, may be required. Often, if minor, these complex clefts can be resolved with local flaps. If major lateral nasal clefts are present, a combination of a forehead flap with tissue expansion and cartilaginous reconstruction in a staged fashion may be required (Fig. 35-9). Occasionally, composite grafts, usually from the concha, can be used to reconstruct the lateral ala with excellent color match and function. Clefts limited to the nose can vary in severity and include the upper and lower lateral cartilages, as well as bones in severe cases. Surgical techniques are individualized according to the severity of the cleft.^{1,24,25} Recognizing the deficiency of soft tissues and often bone in three dimensions can facilitate surgical planning and reconstruction (Fig. 35-10).



Fig. 35-9 **A**, This child illustrates the more “typical” type of Tessier clefts in combination with atypical clefts and hypoplasia resulting from amniotic band syndrome. **B**, Markings for surgery when the child was 13 months old, after tissue expansion, showing the initially proposed combination of advancement flaps, Z-plasties, and forehead flaps, in addition to planned repair of the lip and palate clefts. (Earlier surgery was delayed because of neurologic issues and delayed scalp healing.) **C**, Result at 1½ years of age after the outlined single surgery, along with conformer expansion of the reconstructed eye socket and placement of an ocular prosthesis. **D**, Ten years after repair, awaiting further reconstruction.



Fig. 35-10 **A**, Median facial cleft seen before correction. **B** and **C**, On dissection of the duplicated nasal structures and cranial base, a paired cartilaginous structure was dissected free, which was split in a manner that allowed the superior leaves to be turned upward and create separation of the cranial base; the inferior leaves were turned to create both the nasal floor and cartilaginous septum. The soft tissues were draped over these structures to provide vascular lining. **D**, Next, the lip and soft tissues of the nose and midface were repaired and the cranial vault reconstruction completed. **E**, At 1 year after the initial repair, there was still inadequate soft tissue in the columella, and both lip and midline nasal tissue were shifted to provide the additional tissue. **F-I**, At 1½ years after the initial repair, the added soft tissue of the columella was augmented both with a modified Tajima columellar lengthening, and combined costal and conchal cartilage grafts.

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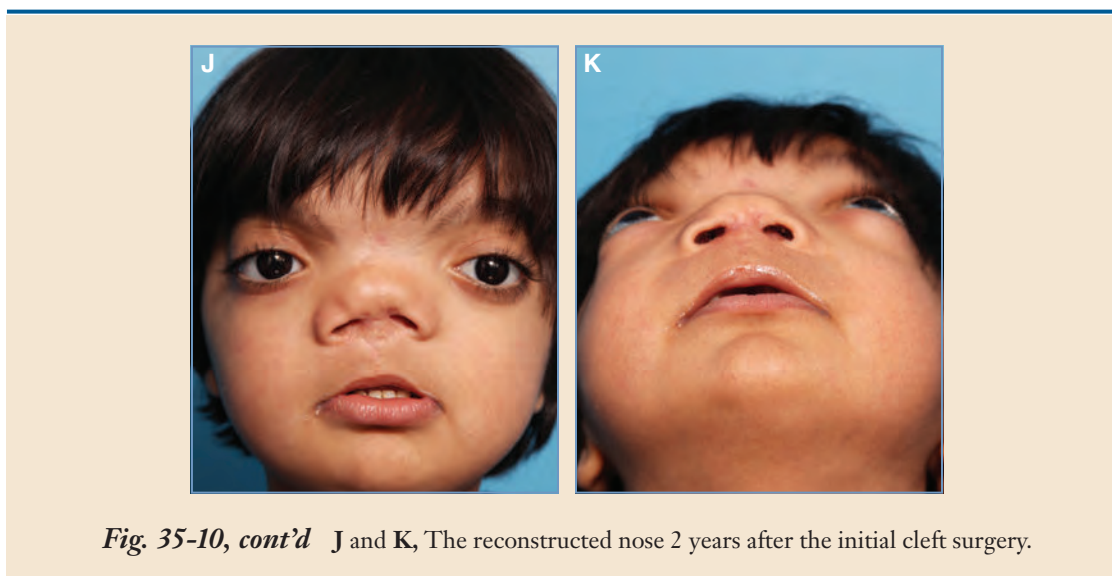


Fig. 35-10, cont'd J and K, The reconstructed nose 2 years after the initial cleft surgery.

Nasal Masses

Nasal masses in the pediatric population have various causes, and proper diagnosis is essential to optimal treatment. The differential diagnosis includes nasal dermoids, gliomas, encephaloceles, and vascular lesions (hemangioma and vascular malformation). The embryologic development of the frontonasal region helps to explain how dermoids, gliomas, and encephaloceles represent a continuum in the disrupted process.^{11,26-30} Early in the developmental process, there is a potential space between the developing frontonasal bony structures and nasal capsule, which develop the nasal cartilages. This prenasal space represents an embryonic “fault line” from the foramen cecum to the skin surface (Fig. 35-11), as clearly shown in the classic paper by Sessions.³¹

Physical examination can usually but not always narrow the differential diagnosis. An MRI with contrast and often a complementary CT scan may be necessary to determine the extent of a lesion. MRI has the advantage of not irradiating the radiosensitive tissues of the eye and is therefore the preferred diagnostic method. The embryologic development of the previously discussed frontonasal region helps to explain the basis for dermoids, gliomas, and encephaloceles^{11,26-31} (Fig. 35-12; see also Fig. 35-11).

Nasal Dermoids

The presence of a frontonasal dermoid may be noted at birth or in early infancy as a midline pore, possibly with protruding hair and possibly with associated widening of the nasal dorsum¹¹ (Figs. 35-13 through 35-16). Nasal dermoids represent the most common congenital nasal mass. Some children may initially present with an infection in the cyst, with swelling and erythema anywhere from the glabella to the nasal tip (see Figs. 35-15 and 35-16). On rare occasions, a child may present with a brain abscess without previous recognition of the cyst or sinus opening from which the infection arose³²⁻³⁴ (see Fig. 35-14).

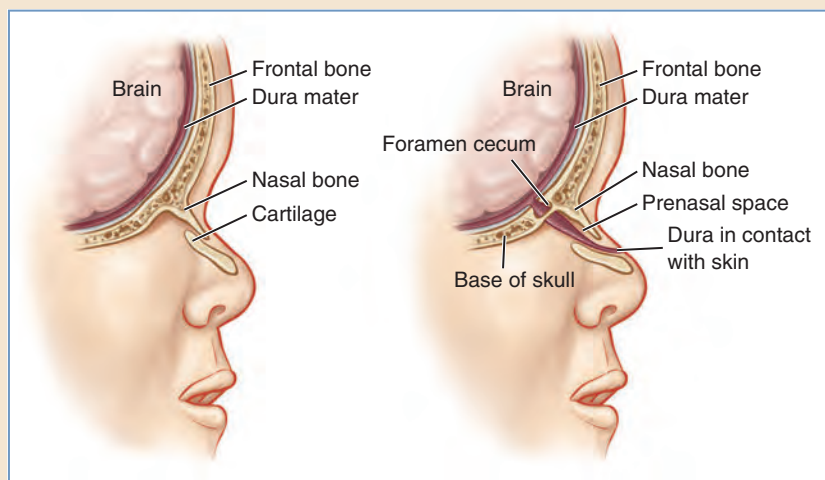


Fig. 35-11 The frontonasal structures develop with an embryonic “fault line” through the area of the prenasal space. The weak points are through the area of the foramen cecum and the fonticulus nasofrontalis, which lies at the frontonasal suture.

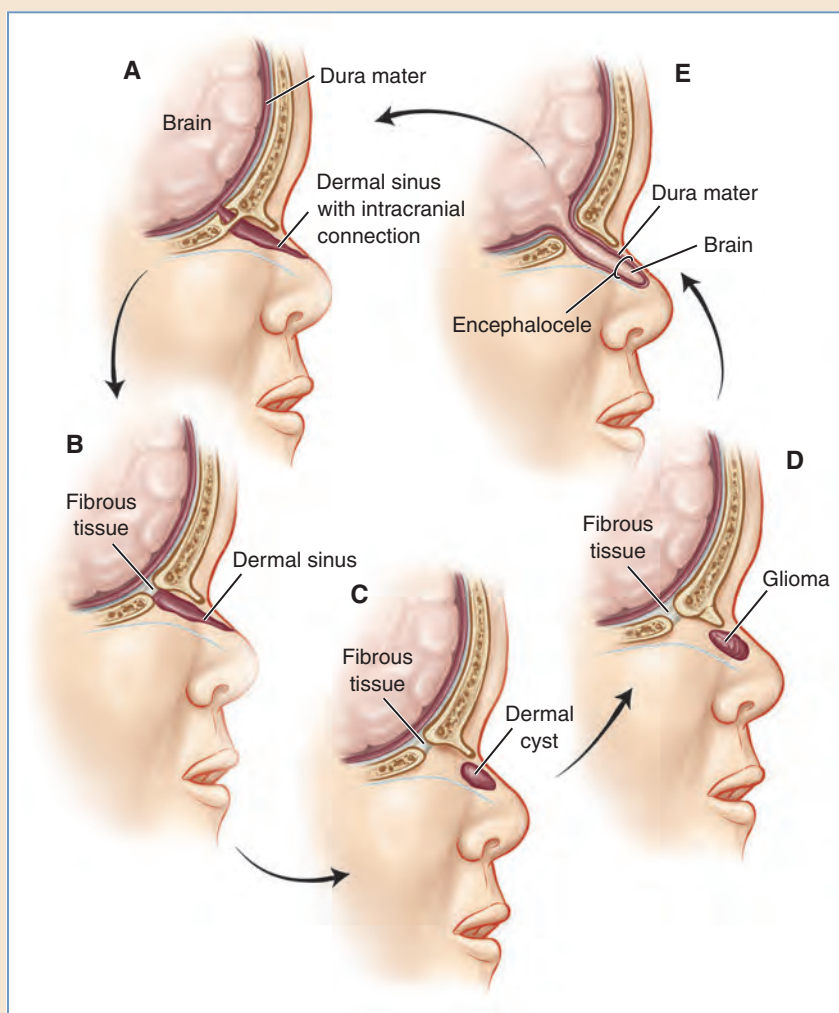


Fig. 35-12 Spectrum of nasal cysts and masses. Classification depends on location and what tissue remnants are left behind. A, Dermal sinus with intracranial extension. B, Dermal sinus tapering to fibrous tract that extends through the foramen cecum. C, Dermal cyst alone. D, Glioma. E, Encephalocele.

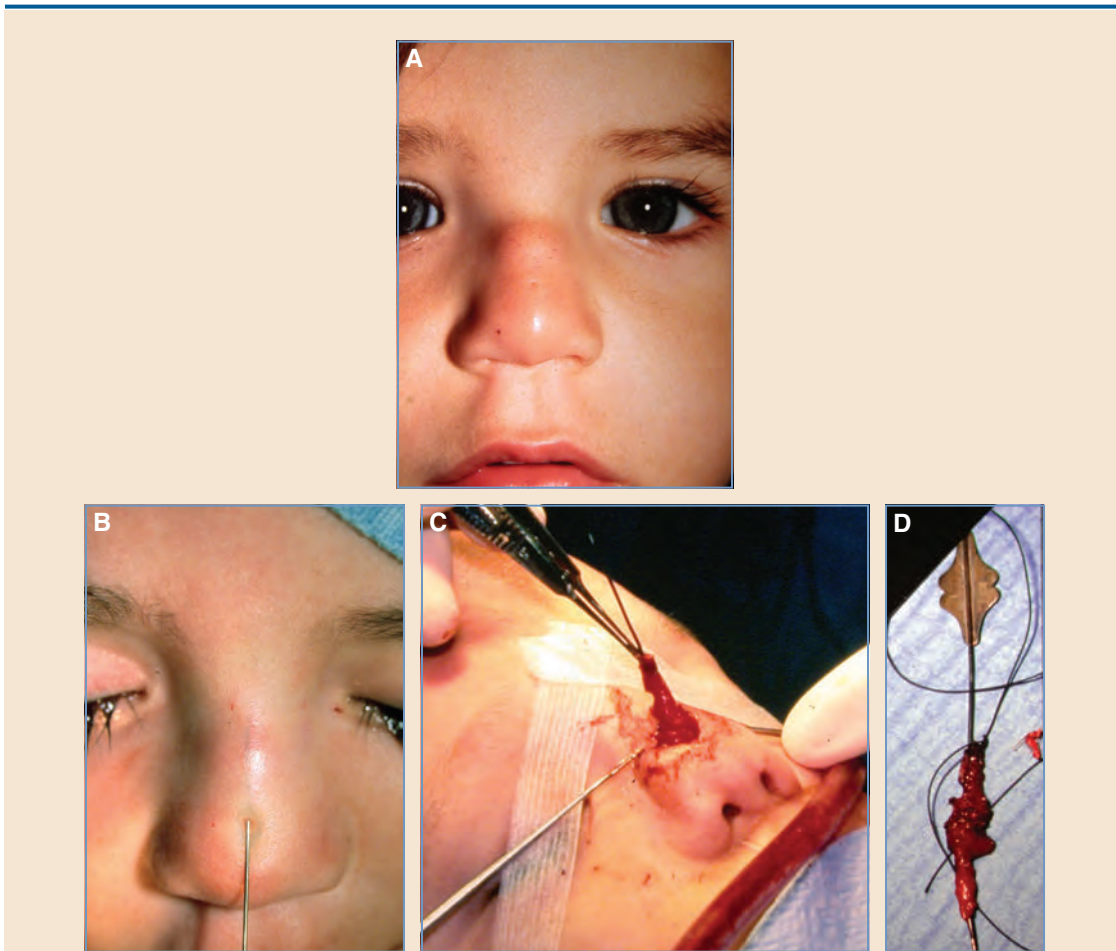


Fig. 35-13 A, This 2-year-old child had obvious swelling and a palpable cyst on the nasal dorsum with an overlying sinus opening. B, The tract was cannulated with a lacrimal probe at the time of excision. C and D, The cyst and sinus before final resection, and fully excised overlying the lacrimal probe.

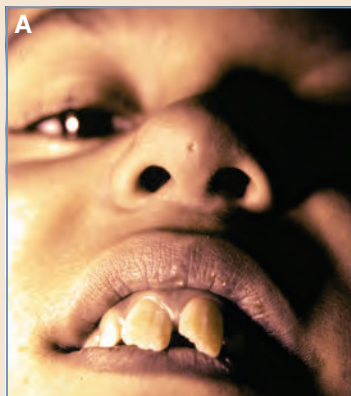


Fig. 35-14 A, This 6-year-old boy was first seen with an intracranial abscess. On close inspection of his nose, a sinus opening was detected at the nasal tip.

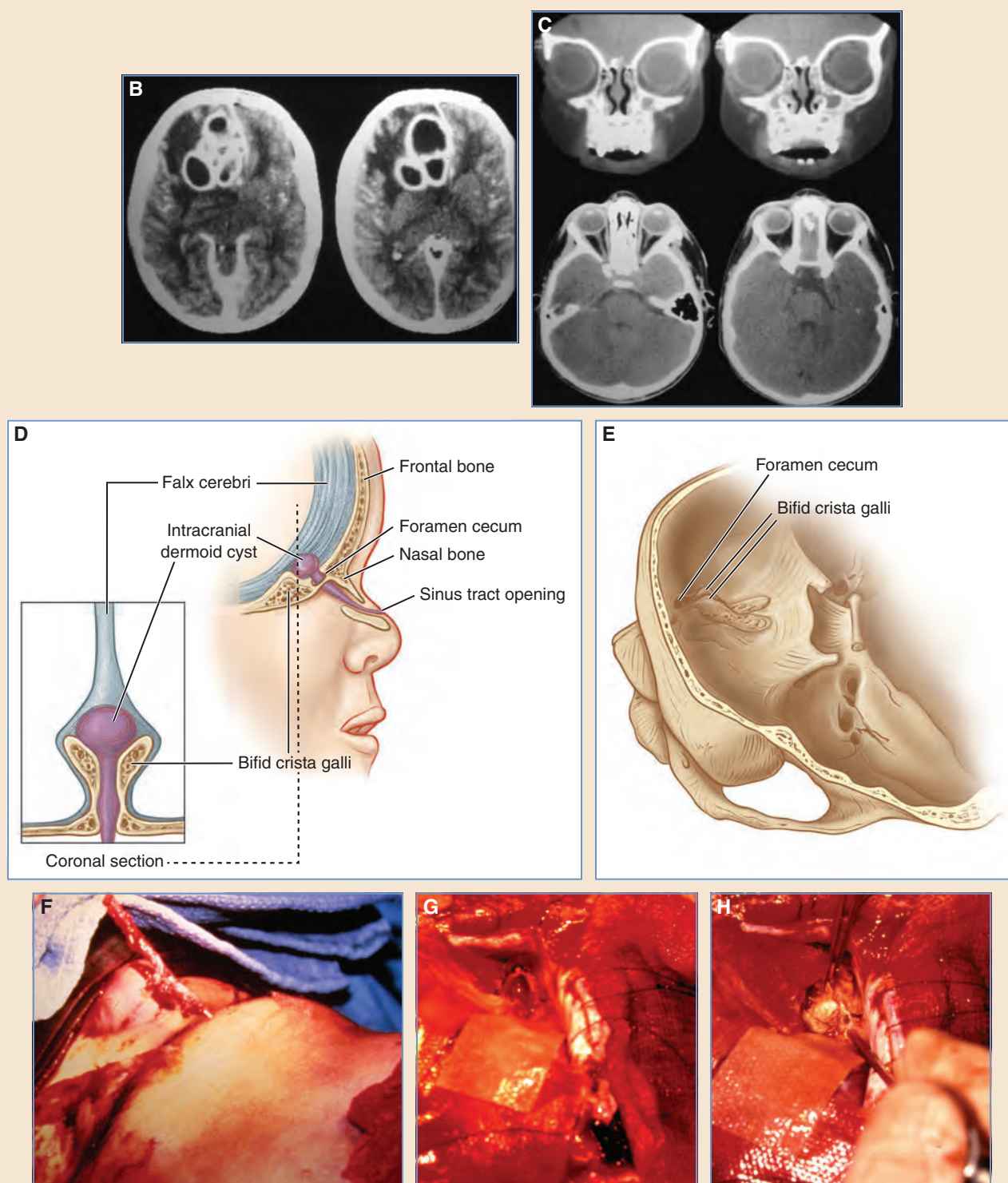


Fig. 35-14, cont'd **B** and **C**, Coronal and axial views show the defect at the level of the foramen cecum. **D** and **E**, Anatomy of the cyst in both sagittal and three-dimensional views of the cup-shaped foramen cecum, widened by the presence of the intracranial portion of the dermoid cyst. **F**, The full length of the extracranial tract is shown. **G** and **H**, Intraoperative intracranial view of the dermoid cyst as resected from between the leaves of the falx cerebri and cup-shaped bony defect illustrated in **E**.

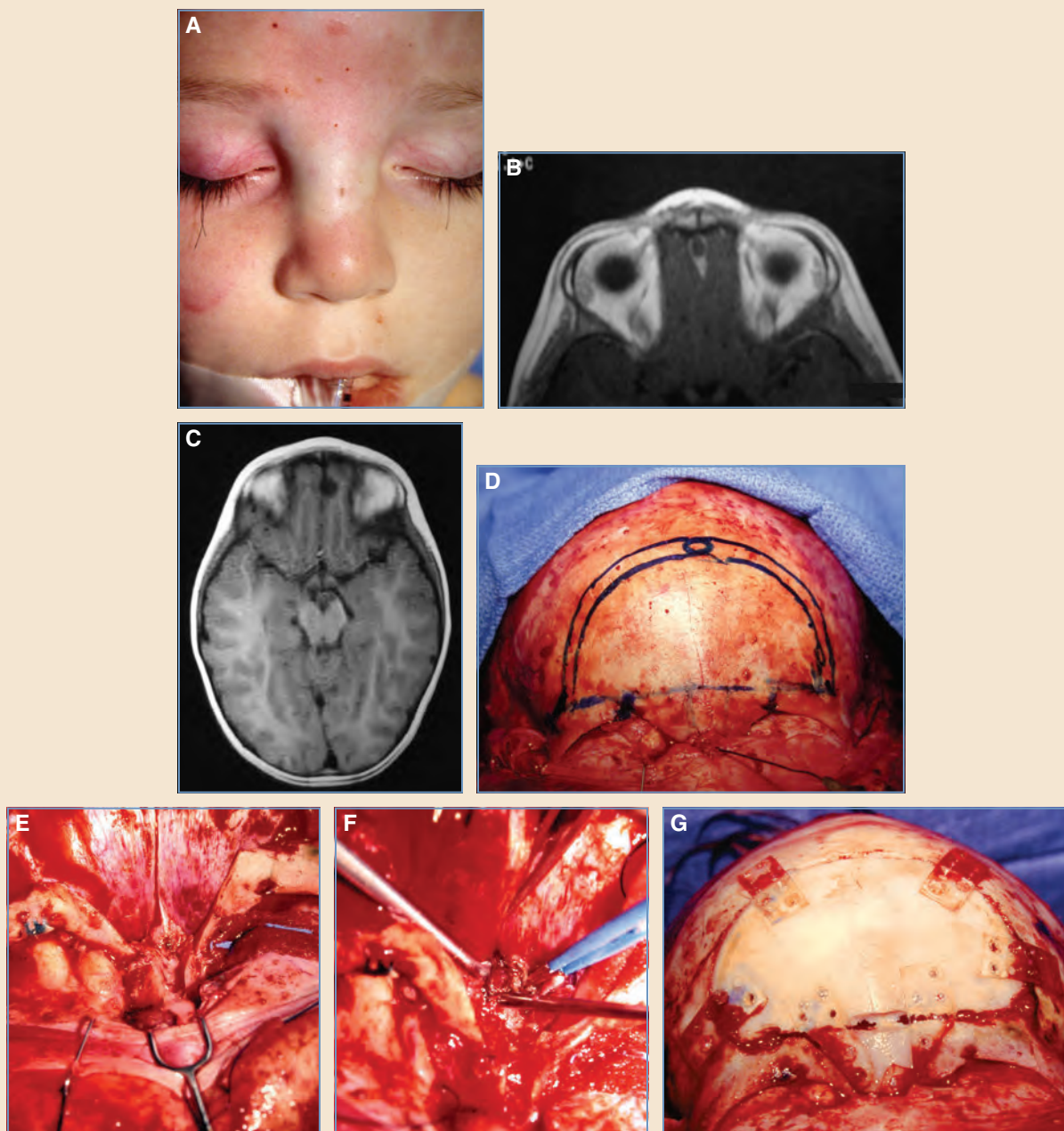


Fig. 35-15 **A**, This unusual dermal cyst and sinus extended not only intracranially but also intraparenchymally (into the frontal lobe). A small hair can be seen protruding from the sinus opening. **B**, MRI showing a cup-shaped defect in the crista galli. **C**, Intraparenchymal location of the cyst in another axial view. **D**, The “keyhole” frontal craniotomy is outlined, along with the planned osteotomy line for removing the frontonasal complex en bloc. **E** and **F**, Excellent views of the area between the falx cerebri, which provides access for resecting the portion of the dermoid within the frontal lobe. **G**, The frontonasal bony complex and frontal bone are replaced and fixed with absorbable plates.



Fig. 35-16 **A**, This infant had intermittent drainage from a sinus opening in the area of the glabella. Despite being given recommendations to proceed with resection, the family did not return for 4 years. **B**, Series of axial views through the area of the cyst, which had eroded the surrounding bone. The films were interpreted as showing long-term osteomyelitis. **C** and **D**, At the time of resection, the ragged edges of the bony defect were clearly visible.

Because the extent of soft tissue tracts may not be recognized beforehand, a CT scan or MRI is essential before surgery. The preference for one study over the other is mostly an individual one, and some lesions may require both studies to determine the continuity of the sinus tract through the foramen cecum and any presence of an intracranial component to the lesion.^{27,28} An MRI scan can provide valuable information regarding the soft tissue tracts associated with dermoids and help to differentiate normal anatomic variants of the anterior cranial fossa from sinus tracts penetrating intracranially (see Fig. 35-15, *B* and *C*). CT scans can provide the best picture of the bony anatomy of the nose and cranial base (see Figs. 35-14, *B* and *C*, and 35-16, *B*). CT scan data can even be translated into a life-sized anatomic replica of the patient's skull. These three-dimensional models can be a valuable planning and clinical adjunct if bony resection

and/or reconstruction is necessary. Lesions with intracranial extension either course beneath the nasal bones through a widened septum and then through the foramen cecum, or through a widened frontonasal suture and then the foramen cecum. In both cases, a bifid crista galli is usually noted on the CT scan, and an intracranial portion of the dermoid may lie between the leaves of the falx. Excision of these more complex lesions is facilitated by a cooperative effort between the plastic surgeon and neurosurgeon using a craniofacial approach.^{35,36}

The surgical approach to “simple” cysts or sinus and localized cyst alone varies, depending on the exact location on the nose or glabella. As previously described, the sinus opening at the base of the columella extends to the nasal spine only, and if associated with a cyst, the sinus is circumscribed for excision, and the remaining dissection of the cyst is accomplished through the labial sulcus. For the remaining sinuses and cysts from the nasal tip to the radix, one should choose the technique that allows the best view of the tract and its final point of attachment, with every effort to minimize the final scar (see Fig. 35-13). Some prefer excision of the sinus opening in combination with an open rhinoplasty approach³⁷; however, if the tract extends for any distance beneath the nasal bones, it may be very difficult to see. A poor field of vision during the first surgery may result in the problem recurring, so every effort should be made to remove the deep component during surgery. If the cyst or sinus is intimately attached to the skin, the tract may be difficult to obliterate if it is not excised directly, which may lead to recurrence. Sinuses and cysts of the glabellar area (fonticulus nasofrontalis origin) without intracranial extension may be approached through a curvilinear incision within the shadow of the lateral nose alone or in combination with excision of the sinus opening. This yields a barely visible scar and gives an excellent view of the area. A transnasal, endoscopically assisted approach has been reported for the resection of lesions that do not have an intracranial component.

For lesions with suspected or confirmed intracranial extension (see Figs. 35-14 and 35-15), excision of the sinus opening begins from the external approach combined with a frontal craniotomy, with delivery of the tract through to the fully exposed frontonasal area, following it through to its intracranial terminus.³⁸ For tracts that seem to terminate in a foramen in the bone, a biopsy will confirm the presence or absence of epidermal elements in the tract. If confirmed, a limited craniotomy is performed for access. For more complex lesions and those with confirmed intracranial extension, the previously described approach is combined with an en bloc removal of the frontonasal bone with direct visualization of the tract, crista galli, and falx through a keyhole osteotomy^{39,40} (see Fig. 35-15). Immediate reconstruction of the nasal and cranial defects is preferred. For patients who have had a significant amount of the bony and cartilaginous skeleton of the nose resected, the nasal dorsum should be reconstructed with a combination of cartilage and bone grafts. If the cartilaginous defect is small, a conchal graft may suffice, but if the defect is larger, a costal cartilage graft is preferred. Most defects resulting from a keyhole resection involve the frontal bone and anterior base of the skull. Cranial bone grafts are optimal for reconstruction and are usually obtained from the parietal bone. Preserving a large, anteriorly based pericranial flap allows its interposition between the dura and reconstructed anterior fossa, separating the nasal contents from the dura.

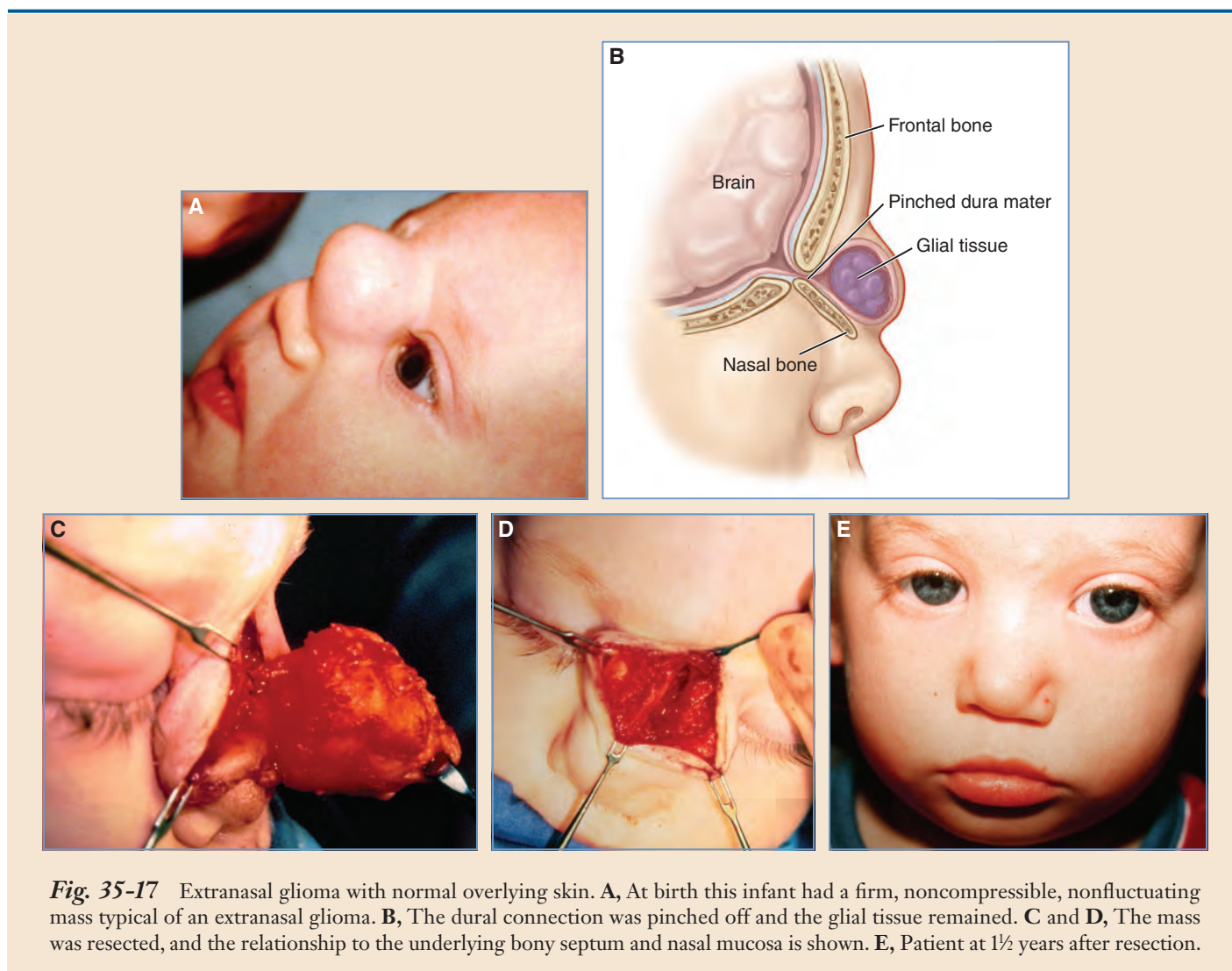
Donor defects are created if the skull is too thin to split. These defects are behind the hairline and can be repaired satisfactorily with allograft and demineralized bone matrix paste. Because these lesions typically occur in the infant or pediatric age group, nasal and frontal sinus development may be affected. Often further nasal reconstruction will be necessary in the teenage years. This is done through an external approach for optimal visibility using autologous cartilage grafts, which can often be obtained from the septum. Frontal sinus hypoplasia can occur after keyhole resection if the frontal sinus origins are disturbed, but this usually does not manifest until adolescence. The external manifestation is a visible depression occurring just above the nasofrontal juncture. This can be corrected with gratifying aesthetic results by using hydroxyapatite cement and a bicoronal approach.

Nasal Gliomas

Nasal gliomas represent heterotrophic neural tissue resulting from deficient regression of neurologic tissue in embryonic development.⁴¹ Gliomas in an extranasal location may still penetrate the bone in the frontonasal suture area and are often associated with both broadening of the nasal root and increased intercanthal distance (Figs. 35-17 and 35-18).

Continued extension through the widened foramen cecum and attachment to the dura in the area of the falx should be ruled out by MRI scan. An MRI scan can distinguish the interface between cartilage, bone, and brain in the region of the frontonasal area. Dural continuity can be ruled out by metrizamide cisternography if the MRI is not definitive.⁴² Gliomas presenting intranasally are usually noted first during physical examination in association with complaints of nasal airway obstruction or evidence of nasal bone distortion resulting from an expanding intranasal lesion¹¹ (Fig. 35-19).

Most often gliomas are confused with hemangiomas.^{41,42} After the diagnosis is made and the extent of the lesion is determined, the lesion can be excised. The surgical approach is again



tailored to allow viewing and excision of the entire lesion. Lesions with an intracranial communication may require a frontal craniotomy for complete excision. Extranasal lesions may require excision of the overlying skin with flap reconstruction when the skin is involved. Endoscopically assisted excision with a modified external rhinoplasty approach may be possible for lesions with a subcutaneous presentation and no intracranial involvement. Histologic examination reveals glial tissue, primarily astrocytes and fibrous tissue; occasionally neurons and ependymal cells are found.³⁷

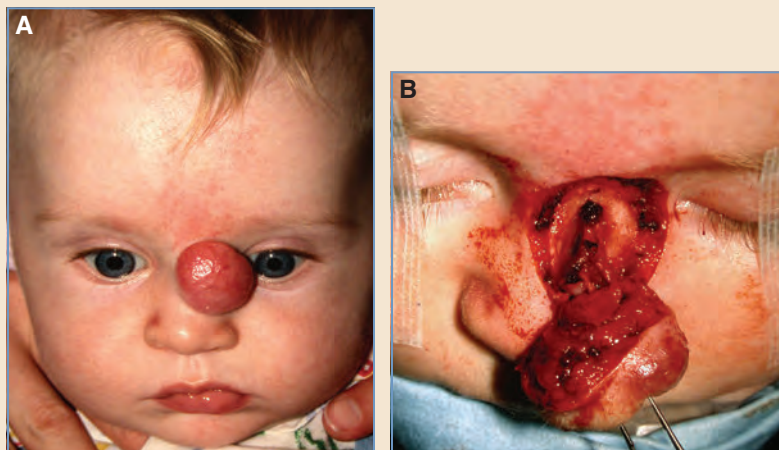


Fig. 35-18 Extranasal glioma involving the overlying skin. **A**, This lesion masqueraded as a vascular lesion, but it was not compressible and did not fluctuate with breathing or a Valsalva maneuver. **B**, Defect in the nasal bone and underlying dorsal septum at the time of resection.

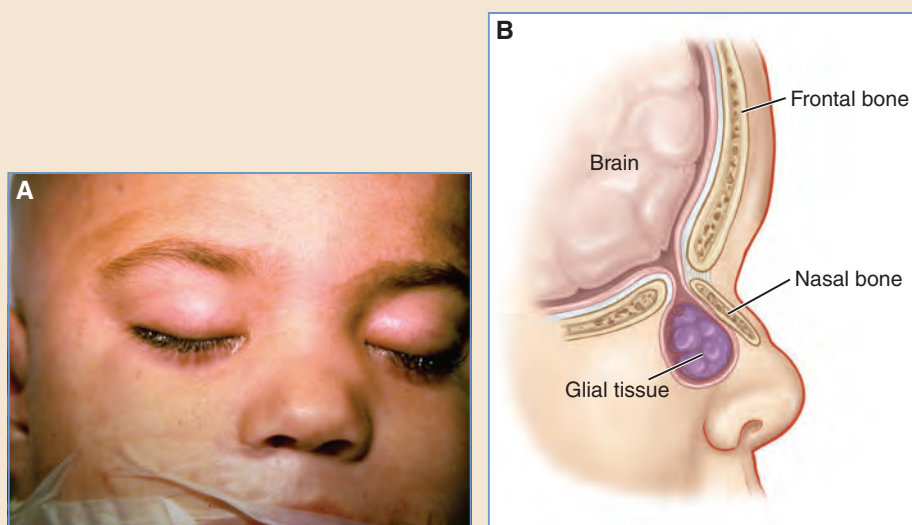


Fig. 35-19 Intranasal glioma. **A**, This 3-year-old child had a gradually enlarging mass in the left nasal cavity that caused splaying of the nasal bone. **B**, Anatomy of the intranasal glioma and its relationship to the overlying nasal bone.

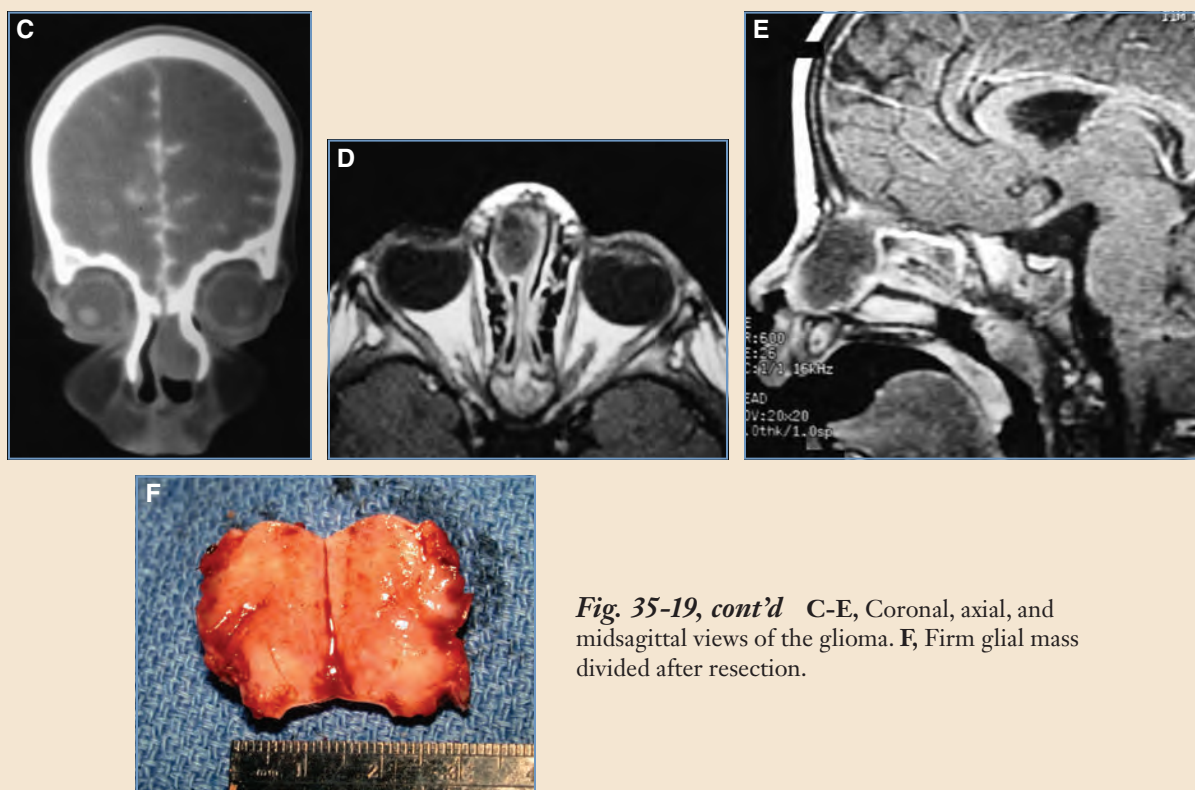


Fig. 35-19, cont'd C-E, Coronal, axial, and midsagittal views of the glioma. F, Firm glial mass divided after resection.

Frontonasal Encephaloceles

Encephaloceles or meningoceles can also present as midline or paramedian nasal masses, usually at the nasofrontal area. A midline or paramedian cystic mass presents at birth and can reach enormous proportions (Fig. 35-20). Size fluctuation may be noted during crying or straining. In general much of the dural sac is filled with cerebrospinal fluid, but it may contain a herniated frontal lobe, along with amorphous glial tissue. Again, the most common path of herniation is through the open cranial base at the site of the previous foramen cecum and the widely patent frontonasal suture.^{15,16} Herniated tissue may extend well into the ethmoid air cells, in which an intranasal component is present. Encephaloceles may be associated with a widened intracanthal distance or true hypertelorism. MRI and MR angiography can give a detailed multiplanar image of the extent of the lesion, the vascular anatomy, and the presence of neural tissue in the herniated sac.²¹ Arteriography is often beneficial to determine the surgical approach and presence or absence of functional brain tissue. Significant defects of the nasofrontal area and anterior base of the skull may be present. A CT scan with three-dimensional reconstruction can give a complete picture of the extent and shape of the defect. A three-dimensional acrylic model, which may be sterilized for operative use, can provide valuable information for planning and executing the bony reconstruction. Treatment is best planned as a cooperative neurosurgical and plastic surgical effort. Histologically, the sac is lined by arachnoid tissue with possible heterotrophic glial tissue, which are accompanied by underlying bony and dural defects. Repair of both the dura and underlying bony defect, usually with cranial bone grafts, is recommended. Bone grafts can be cut

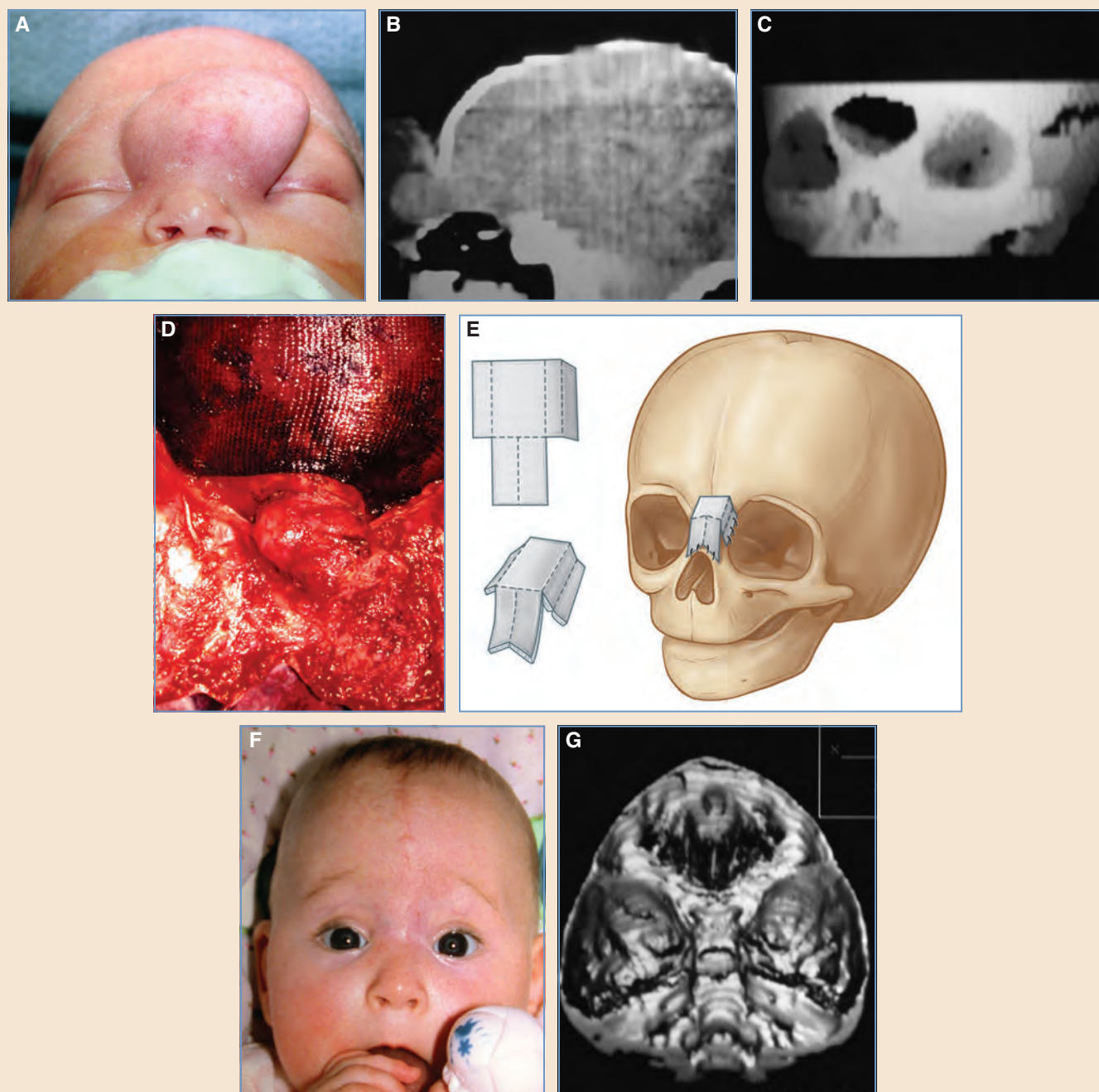


Fig. 35-20 **A**, Frontonasal encephalocele in a newborn infant. **B**, Midsagittal CT view shows a defect in the area of the previous fonticulus nasofrontalis. The continuity of the dura is also shown. **C**, The defect is shown on a three-dimensional CT scan. **D**, Intraoperative view of the encephalocele after it was fully exposed and before resection. **E**, A cranial bone graft was used to reconstruct both the anterior cranial base and nasal dorsum. **F**, Six months after repair. **G**, At 4 years of age, the child showed a trigonocephalic deformity with a prominent midfrontal ridge and antimongoloid slant to the orbits. This CT scan was obtained before craniofacial reconstruction with frontoorbital repositioning and frontal cranioplasty.



Fig. 35-20, cont'd H-J, One year after reconstruction.

and shaped with the three-dimensional representation. Frontal encephaloceles can often result in hypertelorism because of their extension into the ethmoid air cells. Such a residual deformity may require secondary correction, depending on the final aesthetic and functional outcome of the primary repair.

Nasal Hemangiomas

Nasal hemangiomas are one of the most common pediatric nasal masses. They may present as an external red or purple mass, but occasionally they can be entirely subcutaneous, presenting as a swollen nasal tip (see Figs. 35-21 through 35-25). They may have an intranasal component that can compromise nasal airway function. Although some spontaneous involution may eventually occur, nearly all hemangiomas of the nose require treatment to minimize long-term sequelae.⁴³ Physical examination is often sufficient to differentiate hemangiomas from arteriovenous malformations. These malformations are usually present at birth, may have a thrill when auscultated, and are high flow. Purely venous malformations are typically darker than hemangiomas and can be compressed. If a question persists regarding the exact nature of a lesion, an MR angiographic scan can resolve it in almost all cases. Strategic planning for approaching nasal hemangiomas should include minimizing external skin involvement, preserving function, and minimizing long-term distortion of the cartilaginous nasal skeleton.

Several approaches can be used to treat nasal hemangiomas successfully, including serial excisions, steroid injections, external laser treatment, intralesional laser treatments, and resection through an external rhinoplasty approach.⁴³⁻⁴⁶ We use a combination of these therapeutic options based on each patient's individual needs (Figs. 35-21 through 35-23). If a lesion is entirely subcutaneous with normal skin, significant shrinkage can be achieved through the use of intralesional potassium-titanyl-phosphate (KTP) or YAG lasers. This treatment is carried out every 4 weeks for a total of 3 to 6 treatments. A small amount (0.5 ml) of triamcinolone (Kenalog) 5% is injected into the deepest part of the lesion during each treatment. This small amount has not been found to cause subcutaneous thinning, and it helps to decrease proliferation of the hemangioma and to decrease scar tissue while the hemangioma becomes more fibrotic,^{43,46} which significantly

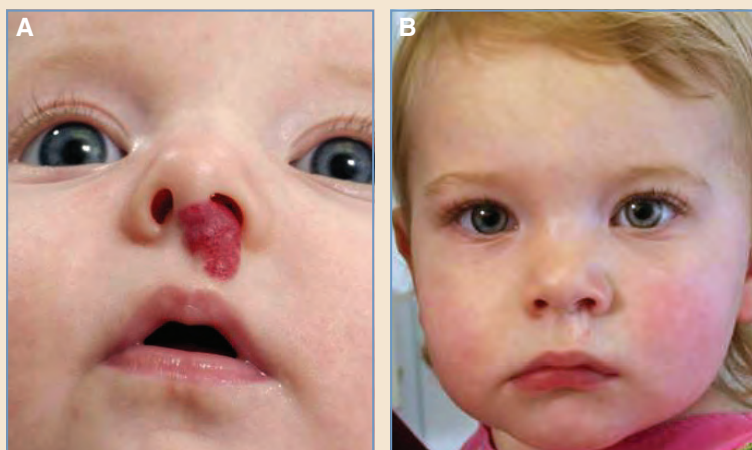


Fig. 35-21 A, This infant had a nasal hemangioma with an internal component causing partial airway obstruction. B, Appearance after serial treatments with a KTP laser, a Candela laser, and steroid injections.

Fig. 35-22 A, This infant had a nasal tip hemangioma with external and internal components. B, Appearance after sequential treatments with a KTP laser, a Candela laser, and steroid injections, and a subsequent external rhinoplasty approach. The residual hemangioma has been debulked and the nasal tip has been reconstructed.



Fig. 35-23 A, Large nasal dorsal hemangioma interfering with the right medial visual field. B, After initial treatment with KTP laser, Candela laser, and steroid injections to decrease the size of the lesion, the residual lesion was treated surgically with debulking and local flap reconstruction.

decreases the vascularity and size of the lesion. If any excess bulk remains 6 to 9 months after the final laser treatment, and there has been no gross regrowth of the hemangioma, an external rhinoplasty approach can be used to remove any residual mass effect.⁴³ A gull-wing incision that does not violate the nasal tip subunit allows excellent exposure to debulk the residual fibrofatty tissue and address the associated cartilaginous deformity. It also allows limited skin reduction, again without violating the subunits.

Often the medial crura of the lower lateral cartilages are splayed in a bifid appearance by the pressure of the hemangioma (Fig. 35-24). This can be reconstructed during an external rhinoplasty by simply suturing from the footplates to the domes, thus reversing the pressure changes



Fig. 35-24 A and B, This child, at 2 years 4 months of age, who had previously received steroid injections and laser treatment for hemangioma of the nasal tip, shows the typical Cyrano nasal tip deformity that follows a bulky hemangioma confined to the nasal tip. Excess fibrofatty and vascular tissue remain, causing the domes of the alar cartilages to splay apart. C, The initial resection was carried out through a gull-wing incision. The upper border of the resection line was estimated but was refined after the fibrofatty vascular mass was resected and the skin redraped. D, Result 1½ years postoperatively with residual vascular changes in the skin. E, Patient at 6 years of age, 4 years after resection.

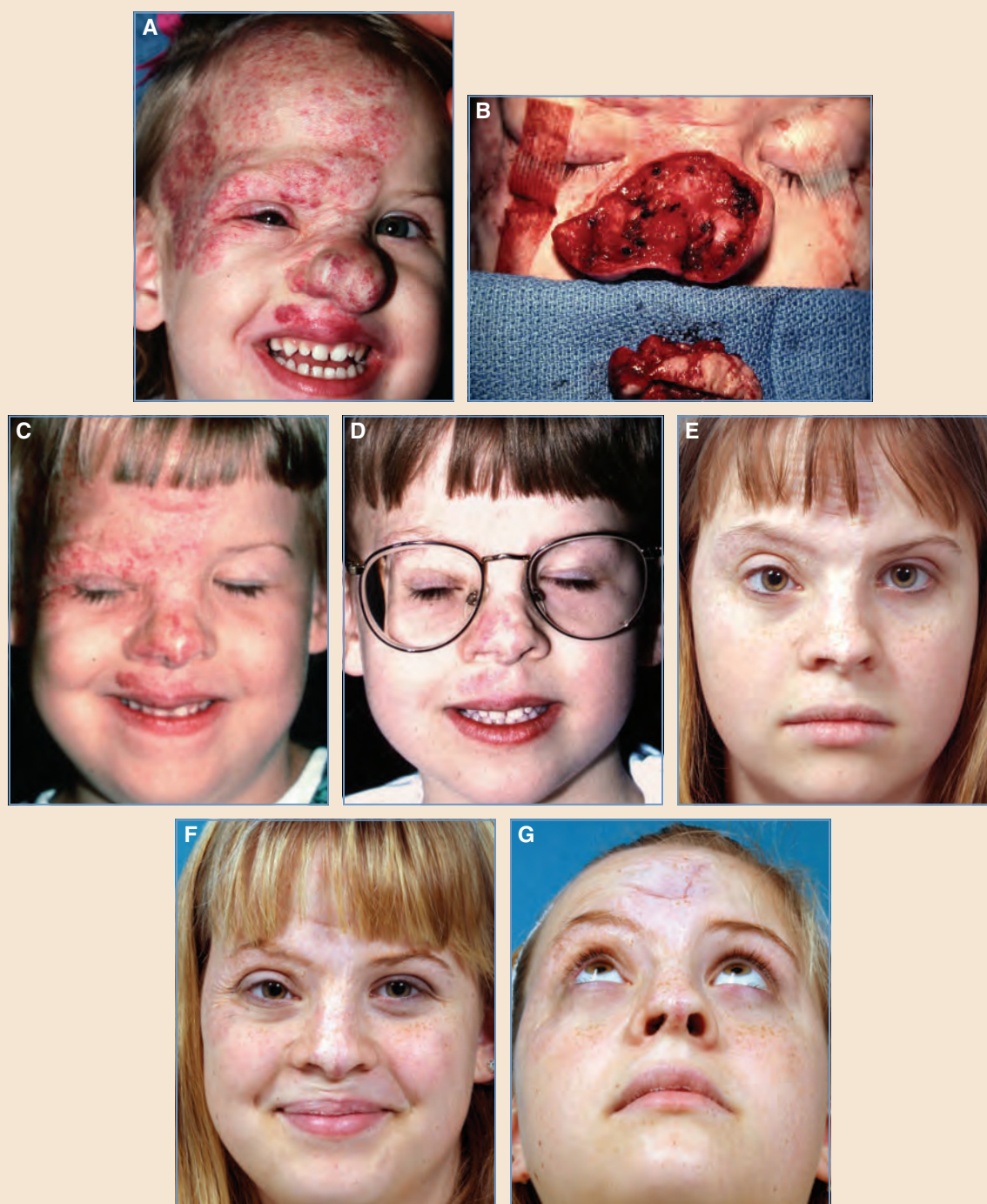


Fig. 35-25 **A**, This infant had an extensive facial hemangioma with numerous ulcerations and significant deformation of the underlying alar cartilages. **B**, At 4 years of age, she underwent extensive debulking, partial skin resection, and repositioning of the distorted alar cartilages. **C**, Patient 3 months after a second debulking procedure and 2 years after the excision shown in **B**. **D** and **E**, Although the residual lesion involuted, the scars matured further, and the nose was allowed to grow. The child is shown at 7 and 16 years of age after the initial resection. **F** and **G**, The patient is shown 8 months after revision of the alar margin, with the addition of a small conchal cartilage graft.

of the hemangioma. If the hemangioma has significant skin involvement, or if the nasal tip skin has ulcerated during the most active proliferative stage, leaving permanent tip scarring, an external skin excision is necessary and may be preferable (Fig. 35-25). Often significant shrinkage of the lesion, as well as salvage of affected skin, can be achieved through a combination of external Candela laser treatment and internal KTP laser treatment. These treatments can be combined with local steroid injection to halt growth, minimize cutaneous involvement, and greatly facilitate the process of serial excision. After maximal shrinkage of the lesion has been achieved, an external excision with a serial approach is advocated. Wide undermining of the normal skin can be achieved, and this skin can be used to replace the involved hemangioma skin over a period of years. Serial excisions are combined with reapproximation of the medial crura, as previously described. In addition, reconstruction may require conchal cartilage grafts when cartilage deficiency is also present to give the best overall aesthetic result (see Fig. 35-25). On occasion, tissue loss can be significant enough to require reconstruction more typical for acquired deformities such as columellar loss and tip loss.

Cutaneous Lesions: Nonvascular

The range of congenital and acquired cutaneous lesions in the pediatric population is covered in Chapter 8. The smaller acquired lesions (0.5 cm or less) present the same issues and concerns as do acquired lesions in the adult population; however, congenital nevi, both melanocytic and sebaceous, may create particular concerns both in their extent and distribution⁴⁷⁻⁵⁰ (Figs. 35-26 through 35-29). Unlike reconstruction of acquired malignant lesions in adults, reconstruction of congenital nevi is typically confined to reconstructing the skin even for extensive lesions, because the underlying supporting cartilage and lining tissues are either uninvolved entirely, or the underlying supporting cartilage may be left intact, balancing concerns about potential functional disturbance and the likely low risk of later malignant degeneration.

Congenital Melanocytic Nevus

Congenital nevi of the nose are uncommon and may be confined to the nose alone or as part of a more extensive nevus of the periorbital region, hemiface, or bilateral face. Although the risk of malignant transformation of giant congenital nevi is well established, the risk of malignant transformation of congenital facial nevi appears to be less common; however, the psychological impact on a child and the child's family from a pigmented facial nevus can be profound. Additionally, some nasal nevi can produce a variety of functional problems from significant thickening of the subcutaneous tissues creating a bulk and weight that may compress the ala and secondarily narrow the nostril. For these reasons we advocate excision and reconstruction of these nevi.⁴⁹⁻⁵¹ Although some surgeons elect to delay the treatment of these very visible lesions until later ages, when nasal growth is more complete and some procedures may be easier, treatment delays may have a significant long-term impact on a child trying to deal with constant peer ridicule.

Treatment of Cutaneous Lesions

Staged Excision (With or Without Adjacent Tissue Expansion)

Although one must always design an excision and reconstruction with the aesthetic nasal subunits in mind, and give consideration to recruiting distant forehead tissue to provide coverage, excision of some of the nasal tip subunit, alar margin, and middle dorsum may be treated quite effectively using staged, serial excision, with or without local nasal tissue expansion (see Figs. 35-26 and 35-27). As long as underlying cartilage is left intact, even a limited reduction in nostril size (following the alar margin excisions) may yield excellent long-term outcomes, with only minor scar revisions of balancing procedures necessary near the completion of nasal growth.



Fig. 35-26 A congenital nevus involving the greater part of the alar subunit of the nose in this 11-year-old girl. Although this lesion is typically considered large enough to require a forehead flap or composite graft reconstruction, a staged excision and adjacent flap movement were performed. **A-C**, Three views of the area of involvement, which can be seen extending across the nostril margin but not intranasally. The resection and movement of flaps were done without resection of the cartilage. **D**, Results 2 months after first-stage resection. **E** and **F**, One and a half years after the first excision and 2 months after the second excision, the basal view shows the intermediate thickening of the soft tissues along the nostril margin. **G** and **H**, One year after the final thinning of the soft tissue along the nostril margin; the patient is 16 years of age.

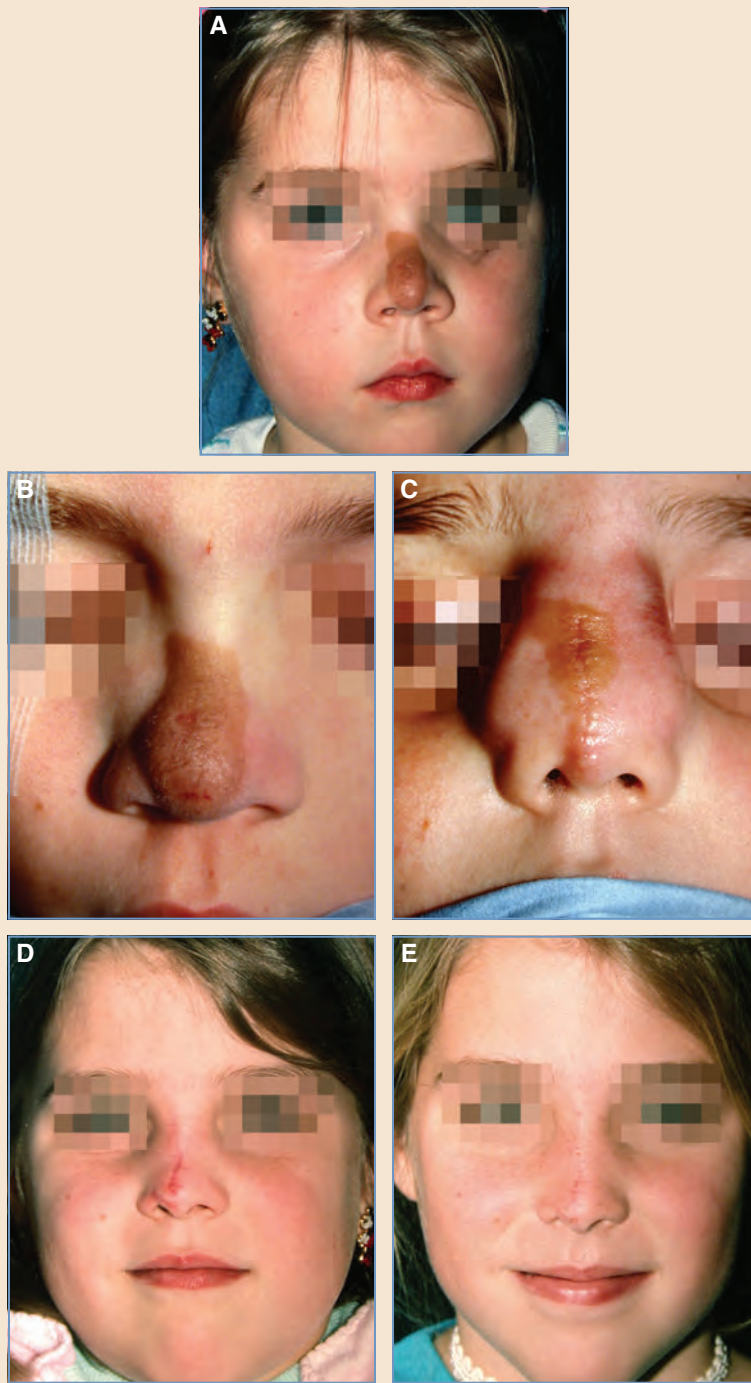


Fig. 35-27 A, This 5-year-old girl had a bulky congenital pigmented nevus of the nasal tip and dorsum. Although excision and reconstruction with a forehead flap was an option, the lesion was excised with expansion of adjacent normal skin. B, Close-up view of the nevus before two 3 cc custom expanders were placed through an incision within the nevus that also allowed a conservative partial resection of the nevus. C, Appearance of the expanders just before resection of the remaining nevus. D, Patient 8 months after resection. E, Same patient at 10 years of age.



Fig. 35-28 Early stages of reconstruction in a child with a congenital giant pigmented nevus of the face. **A**, Patient at 6 months of age. **B**, After successful expansion that began at 1 year of age; with expanders in both the forehead and neck, the forehead flap was oriented so that the donor scar lies along the brow and continues out into the zygomatic temporal region. **C**, Appearance a few days after resection of the nasal nevus and expanded flap reconstruction. **D**, Six months after the procedure shown in **B**, an expander was placed in the child's cheek. After an additional 3 months of expansion, the remaining nevus of the cheek was resected, and the forehead flap was lifted from the nose, thinned, and replaced. Patient is shown 10 days postoperatively. **E-G**, At 10 years of age, she is still undergoing excision of remaining nevus in the eyelid and refining of the soft tissues of the nose and cheek. Final reconstruction is expected when the patient is a teenager.



Fig. 35-29 This infant was born with extensive sebaceous nevi, without associated central nervous system involvement. **A**, Larger lesions covered large segments of her scalp, and she had multiple linear lesions in the central and lateral portions of the face. This type of lesion is approached using a combination of tissue expansion and staged excisions of some of the narrower linear lesions. These excisions may be timed to allow partial excision at the time of expander placement and repeat excisions later at the time of more extensive excision and flap reconstruction. **B** and **C**, The first set of expanders at the completion of expansion. **D**, Patient's appearance 5 years after the start of staged excision and reconstruction, with the greater part of the nevus on the nose, forehead, and scalp excised but residual lesions in the nasolabial region, lip, and lateral eyebrow just before further excision.

Excision With Full-Thickness Skin Graft Coverage

Although nevus excision only involves the excision of skin and subcutaneous fat to the perichondrial or fascial level, full-thickness skin grafts typically do not provide an excellent long-term outcome. However, they may be beneficial for removing deeply pigmented, very visible lesions in children younger than some surgeons may be comfortable with. Although full-thickness skin graft coverage of the nose in combination with a single, large, expanded, full-thickness skin graft to the periorbital area has been used effectively in the past to provide excellent long-term results,⁵¹ we now prefer early coverage with an expanded forehead flap.

Expanded and Nonexpanded Forehead Flaps

Although there are differences of opinion regarding the benefits of expanding the forehead before using a forehead flap for nasal reconstruction, our extensive experience in treating large and giant congenital pigmented nevi of the face has showed unique benefits of the expanded forehead flap (see Fig. 35-28). When advancing the expanded central forehead laterally to excise nevi of the lateral forehead and temporal region, there is a relative “redundancy” of skin in the glabella and the ipsilateral side of the nevus excision. This tissue can be used based on the same supratrochlear vascular pedicle as a standard forehead flap, as a small flap to cover the nasal dorsum alone, or for total nasal coverage, with the added benefit that the donor scar will lie along the brow, rather than centrally in the forehead (see Fig. 35-28, *C* and *D*). Our experience with expanded forehead flaps in the early stages of treating large and giant nevi is still limited (in the 18- to 24-month age range), but our results clearly support its role as a powerful reconstructive option.

Linear Sebaceous Nevi

Linear sebaceous nevi may present in isolation as a lesion or a series of striped lesions on the nose, or as part of a larger more extensive facial sebaceous nevus associated with sebaceous nevus syndrome.⁵² Although some physicians have elected to treat linear sebaceous nevi with lasers alone, any remaining portions of a lesion require an extended series of treatments, and still may be visible enough to create an unsightly deformity. The skin between stripes of lesion is normal, and serial linear excisions can preserve this normal tissue and allow excellent long-term outcomes. With an extensive facial nevus, staged linear excisions of the nasal lesion may be performed during the same surgery by either placing tissue expanders, or removing them and undertaking an extensive excision and flap reconstruction (see Fig. 35-29). Because the latter procedures are typically carried out with 11 to 12 weeks of expansion, with a 4- to 6-month interval between each group of procedures, this allows multiple opportunities to serially excise nasal lesions.⁵²

ACQUIRED NASAL DEFORMITIES

Acquired nasal deformities are most commonly caused by trauma. Traumatic deformities can be divided into those with an open skin component and those of a closed nature. When either open or closed nasal injuries occur, they can result in permanent nasal deformities. If the injury occurs at a young age, these deformities may not become apparent until the child has grown, at which point they may present with a saddlenose deformity or even complete nasal hypoplasia.

Closed nasal trauma injuries are common, are associated with sports or occasionally fisticuffs, and may involve injury to both the cartilaginous and bony components of the nose (Fig. 35-30). The initial evaluation should include a thorough history to reconstruct the mechanism of injury and approximate the force of impact. Standard radiographs may not reveal the extent of injuries in a developing nose, because there is a tendency for the nasal bones to undergo greenstick fracturing rather than shatter. Plain radiographs are not a substitute for a thorough examination. Computerized tomography of the maxilla and nose in both the axial and coronal planes can provide useful clinical information. An intranasal examination is essential to determine whether a septal hematoma is present, which can lead to late pressure necrosis of the septum. A hematoma often presents as a unilateral purple swelling that can completely obstruct the nares. Patients with a septal hematoma often have pain that is disproportionate to the visible extent of their external nasal injury. Treating a closed nasal fracture consists of closed reduction and external splinting. If a septal hematoma is present, it should be drained, and any loose fragments of septum should be debrided.^{51,52} Plain gut is used in a continuous mattress stitch to approximate the septal mucoperichondrial flaps and prevent recurrence of hematoma (Fig. 35-31). An external thermoplastic splint is applied, and no intranasal packing is used. If a septal hematoma is present, a short course



Fig. 35-30 **A**, This 8-year-old girl had a bony and cartilaginous deformity of the nose that occurred when she fell off a horse. **B**, Same patient after closed nasal reduction.

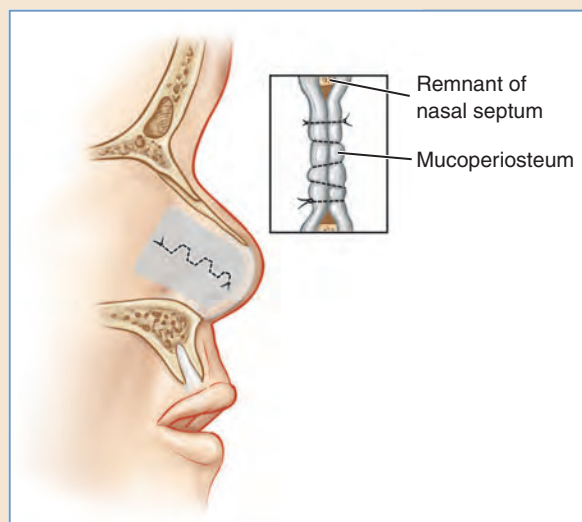


Fig. 35-31 “Weave” technique for avoiding recurrent septal hematoma after evacuation.

of oral antibiotics is prescribed. After 1 week the splint is removed. Contact sports are not allowed for 4 weeks. Open surgery for acute treatment of a closed nasal fracture is not indicated, because the skin and periosteal attachments holding the comminuted nasal bones can be disrupted, making stabilization challenging. Sequelae of untreated nasal fractures with or without septal hematomas include airway obstruction, aesthetic deformity, and saddlenose deformity. Even if optimal treatment is administered, there may be a residual deformity that may become more apparent as facial growth progresses. Parents should be made aware of this possibility during the initial discussion of the injury and required treatment.⁵¹

Open injuries often occur as a result of motor vehicle accidents, falls, animal bites, and sports injuries. Isolated nasal trauma requires thorough intranasal and extranasal examinations. Plain radiographs and CT both can provide valuable information. As with closed nasal trauma,

it is imperative to determine whether there is a septal hematoma with open trauma. Open nasal fractures require reduction, inspection for septal hematomas, and prompt repair of the skin lacerations. CT scanning is recommended to rule out underlying maxillary injuries. If maxillary fractures are also present (and are more significant than nondisplaced greenstick fractures), they are treated with open reduction and resorbable fixation followed by closed reduction of the nasal bone fractures once the skin has been repaired. Wide undermining of the nasal skin should be avoided, because this further destabilizes bony fragments. Only obviously devitalized skin should be debrided, because even macerated traumatized skin is often viable. External splinting is used, and occasionally resorbable nasal packing is used for very unstable fractures. Antibiotics and analgesics are continued for 5 to 7 days. A conservative approach is recommended for the initial treatment of most open nasal fractures, followed by at least 12 months of observation. Approximately 40% of children who have significant nasal fractures exhibit a residual external or intranasal deformity when they reach skeletal maturity. Obstructive symptoms infrequently warrant intervention before skeletal maturity. A conservative approach is also recommended in these patients, with septal scoring and splinting, and turbinate reduction to improve the nasal airway if intervention is necessary before adolescence.⁵¹⁻⁵³ If there is a symptomatic residual deformity at skeletal maturity, it requires an open rhinoplasty approach, often with cartilaginous reconstruction, septal reconstruction, and fracture reposition of the nasal bones. Autologous cartilage is advocated if augmentation is required, especially for the nasal dorsum. Septal cartilage can be useful for small defects, but costal cartilage seems to be more effective for optimal long-term results with larger dorsal defects.

Animal bites are most commonly from canines. Young children are particularly vulnerable, because they are at face height to many dogs. Teenagers may tease or roughhouse with dogs, putting their noses at risk of bite injury. Generally, the initial puncture by a dog's teeth is followed by the victim pulling back, which often results in partial or complete tissue avulsion (Fig. 35-32).

Initial treatment should include debridement, updating tetanus prophylaxis, and oral and topical antibiotics. The first steps of treatment are inspection of wounds with removal of all definitively devitalized tissue and prevention of secondary infection. Given the excellent blood supply in the area, initial debridement should be conservative. After the tissues have demarcated their viability, the residual defect can be evaluated. If there is no significant tissue loss, scar revision may be considered 9 to 12 months after the initial injury, allowing time for scar maturation. If a true tissue avulsion is present, plans for a staged reconstruction should be discussed with the patient and the family. Depending on the defect, local rotation flaps, forehead flaps, or composite grafts may be necessary for repair.⁵⁴ Alar defects can be treated with local rotation flaps combined with conchal grafts or composite grafts for alar airway support. Larger defects of the skin and cartilage may require a forehead flap or nasolabial flaps, depending on the extent of tissue loss.⁵⁴ Tissue expansion may be valuable for obtaining sufficient soft tissue for reconstruction. If adequate soft tissue coverage is available, the underlying nasal framework can be reconstructed at the same time. Significant defects involving the tip, dorsum, and lateral nasal sidewall cartilage are best treated using rib cartilage to construct a framework for supporting the overlying soft tissue flaps. Once the initial framework and soft tissue reconstruction have been accomplished and healed, several refining procedures can be considered. These include scar reduction and debulking procedures, aesthetic tip-plasty, adding conchal cartilage grafts to minor defects, laser treatment of scars, and other modifications. Defects of the columella may require adjacent flaps, composite auricular grafts, or combinations of these procedures (Figs. 35-33 through 35-35). The patient,

the patient's family, and the treating plastic surgeon must all understand that reconstruction for extensive nasal defects will take multiple stages over a period of years. Significant improvement can be expected, but there will be residual donor- and recipient-site scars, and these will be permanent. The animal involved in a bite trauma must be checked for rabies and reported to animal control authorities. Because lawsuits often result from dog bites, careful documentation is essential, along with a conservative estimate of the final aesthetic and functional outcome. It is impossible for the surgeon to estimate the total number of procedures and final outcome after the initial intervention, although many insurers and attorneys demand this information.

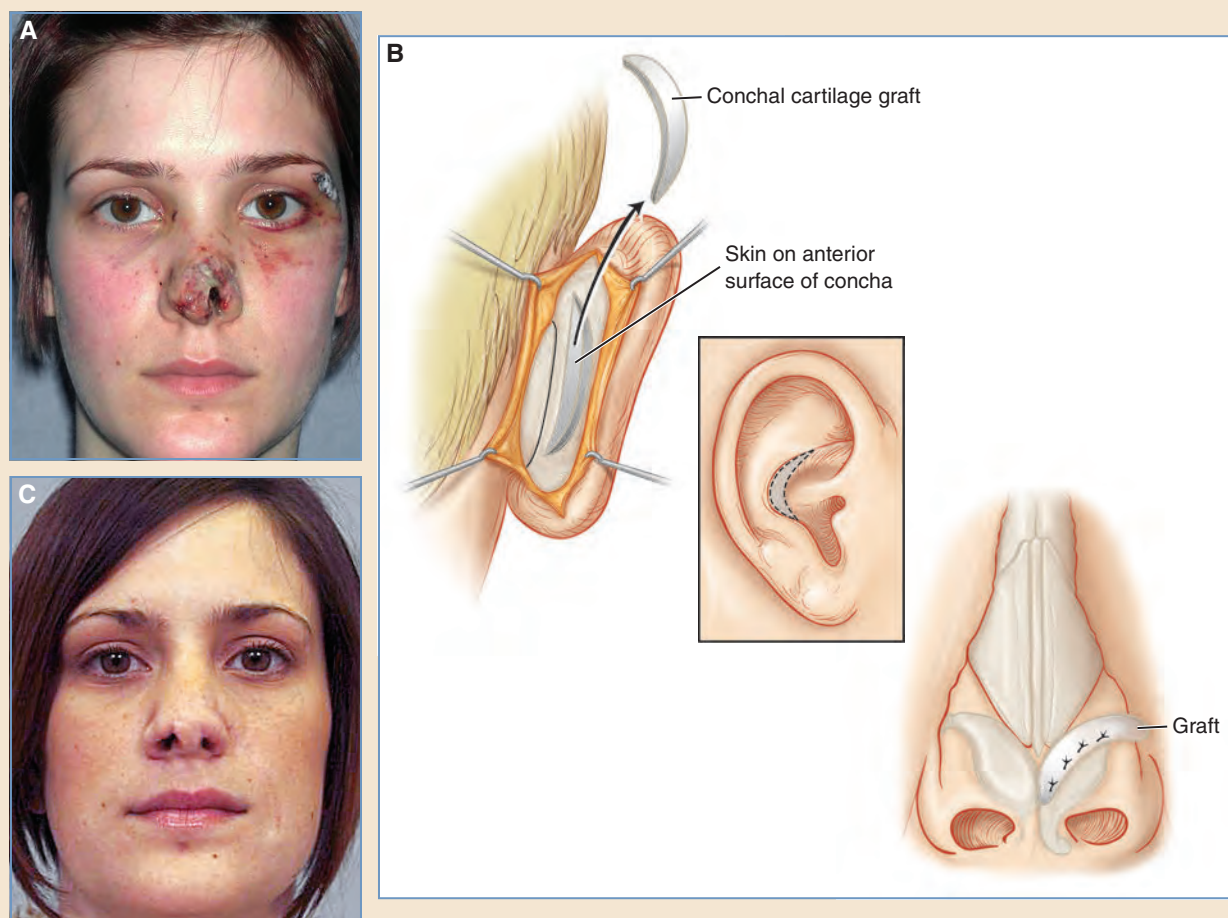


Fig. 35-32 A, This teenager sustained a nasal avulsion injury after she was bitten by a dog. Note the loss of the lower lateral cartilage and caudal upper lateral cartilage with extensive skin loss. B, Technique for harvesting and applying conchal graft for nasal reconstruction. Note that a graft can also be harvested with overlying skin as a composite graft. C, Same patient after staged reconstruction using conchal grafts for cartilaginous nasal vault reconstruction and a left nasolabial flap for external and internal soft tissue reconstruction.



Fig. 35-33 A and B, This 4-year-old boy was seen for reconstruction of columellar and tip defects resulting from partial necrosis of the columella caused by the pressure of the nasal cannulas that were used while he was in the neonatal intensive care. C, The patient returned at 16 years of age after undergoing three operations, including an attempted columellar advancement according to the method of Cronin, two small composite grafts, and a scar revision. These procedures left him with considerable scarring and, according to the patient and his family, a more noticeable deformity. D, Lateral view before further reconstruction shows shortening of the columella, blunting of the nasal tip, and notching of the tip. E, Patient at 17 years of age after reconstruction using an open rhinoplasty approach. After osteotomy and infracture of the nasal bones, septoplasty and septal cartilage graft to the columella, and flap reconstruction using portions of the previous composite grafts for additional tip support, the shape of the columella and the nasal tip was restored. F, Lateral view 1 week after surgery shows improved tip projection and smoother contour of the columella. G and H, Two months after reconstruction. This patient was seen on one other occasion to discuss minor scar revision, but he elected to postpone this.



Fig. 35-34 This baby girl presented with a hemangioma that was rapidly destructive despite medical management. At presentation, she already had necrosis of her columella. **A**, At 13 months of age, a point involution was beginning. She was also diagnosed and underwent surgery at 2 years of age for moyamoya disease, an intracranial vascular malformation. **B** and **C**, At 2 years 9 months, her first surgery involved developing a flap based on the center of the enlarged upper lip, as a vascularized flap turned 180 degrees and inset into the tissue along the border of the central nose. This provided the needed tissue for the subsequent reconstruction of the columella. During the same surgery, the lateral nose and medial canthus and brow tissue were reduced. **D**, Seven months after the first surgery. **E**, Nine months after the first surgery and 1 month after the second surgery, further shaping of the nasal and lip soft tissues was performed. **F**, At 4 years, 6 months of age after the second surgery. **G-I**, At 9 years of age, which was 7 months after further reconstruction of the columella with a flap and conchal cartilage graft. She will be a candidate for additional scar revisions and strut graft in the future.

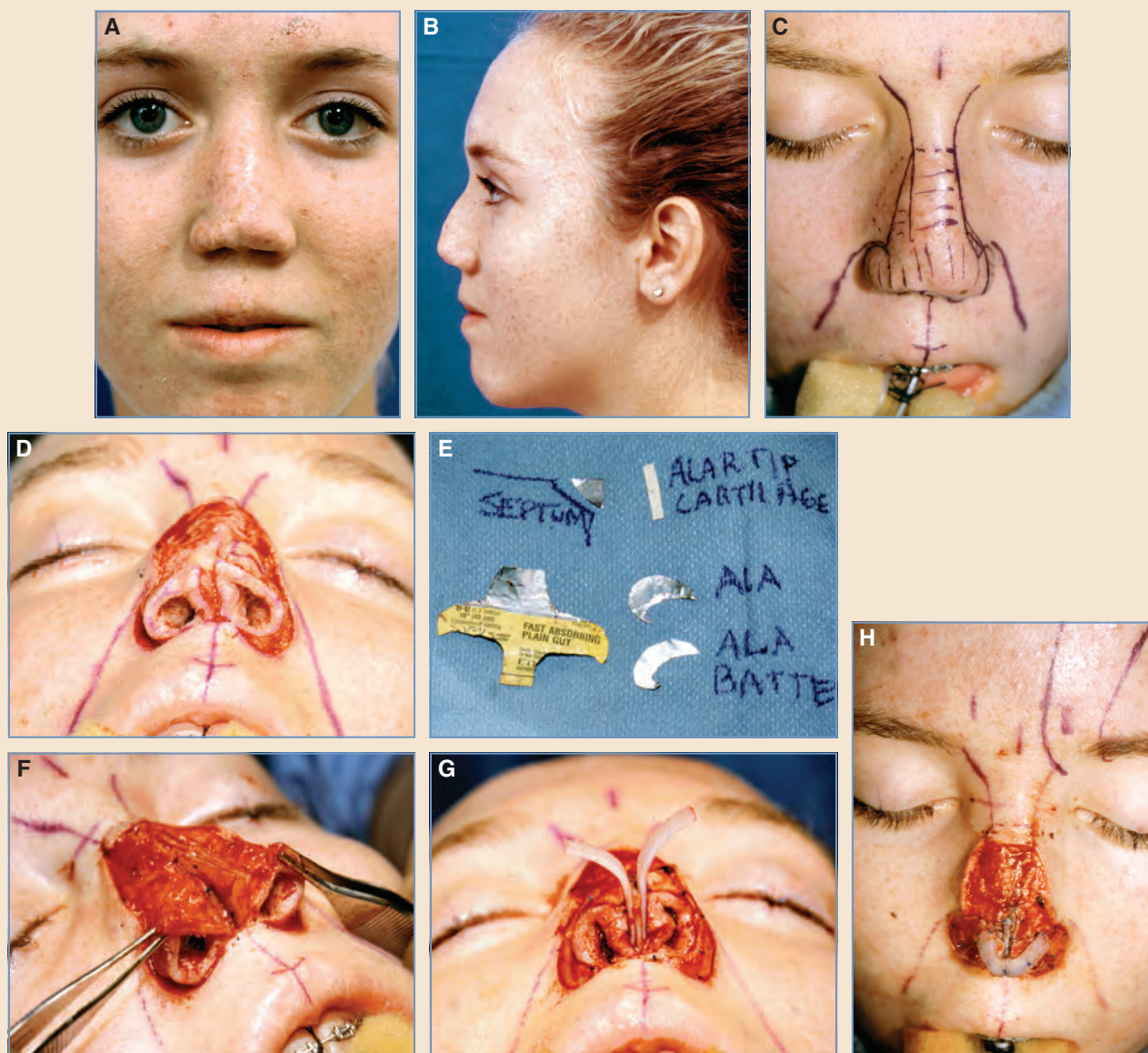


Fig. 35-35 Forehead flap reconstruction of an extensive defect that occurred after tissue destruction resulting from a hemangioma. **A and B**, This 14-year-old girl is shown after destruction of the soft tissues and cartilaginous support of the nasal tip and columella subsequent to an involuting hemangioma. The skin over the entire nose is scarred and atrophic. The columella and right rim had been repaired by means of local tissue rearrangement when she was a young child. **C**, The skin of the nasal units is marked for excision. **D-H**, The angle of the septal cartilage and the entire alar cartilage complex are absent. Templates were made to guide the exact replacement of the missing nasal skin and absent septal angle, tip cartilages, and alar support grafts. A triangle of septal cartilage was fixed to the residual septum with lateral spreader grafts to re-create the septal angle. Septal cartilage strips were fixed to the distal septum and hinged laterally to re-create the medial, middle, and lateral crura to restore support and contour to the distal nose.



Fig. 35-35, cont'd I and J, The nose was resurfaced with a full-thickness forehead flap. K, At 3 weeks, the forehead flap is healed to the recipient site. The forehead donor site, which could not be closed under the hairline, was allowed to heal by secondary intention. L-N, Three weeks after transfer, the forehead skin was re-elevated with up to 3 mm of subcutaneous fat. In this case it was maintained as a bipedicle flap hanging from its origin at the brow and from the distal columella inset. In most instances, the flap is completely elevated and temporarily positioned to the side of the face. The underlying excess of subcutaneous fat and frontalis is exposed. This is excised with a scalpel to re-create the subtle three-dimensional contours of the nasal surface. The flap is resutured to the recipient site with quilting and peripheral sutures. O, The flap was divided 3 weeks later, and its proximal aspect was returned to the forehead as a small inverted V at the brow. The forehead scar was subsequently revised. P-R, Postoperative result at 19 years of age. The excess fullness of the left alar base could be improved by minor revision. (Courtesy of Frederick J. Menick, MD.)

KEY POINTS

- The definitive nasal structures, both external and internal, develop during the fourth to eighth weeks of gestation concomitantly with the developing primary and secondary palate.
- Facial and limb malformations result from deficiency or excess of molecular signaling (such as Sonic Hedgehog, fibroblast growth factor, and retinoic acid signaling). Similar phenotypes, such as clefting, may result from either deficiency of the appropriate midline tissue or excess of other midline tissue so that the appropriate processes cannot meet to fuse in the midline.
- Because the appearance of the nasal septum varies in radiologic examination depending on the patient's age, one must interpret imaging evidence of midline defects and sinus tracts carefully. In fact, 14% of patients younger than 1 year of age have no midline ossification of the anterior fossa or septum.
- One can best understand the spectrum of congenital frontonasal masses (dermoid cysts and sinuses, gliomas, and encephaloceles) with a clear understanding of the embryology of frontonasal development and the paths of potential embryonic "faults" through the foramen cecum and frontonasal suture.
- The choice of radiologic technique to evaluate frontonasal cysts and sinuses varies partly on surgeon preference. Some surgeons think that the bone anatomy and area of the crista galli are better delineated with a CT scan, whereas others think that the sinus tract is better delineated with an MRI. Complex cases with intracranial extension may best be studied with the use of both.
- Dermal cysts and sinuses with obvious intracranial extension require a craniofacial approach typically through a keyhole frontal osteotomy with initial resection of the intracranial dermoid. For patients with indefinite extension, the tract is followed to its tapered stalk, and a biopsy specimen is taken to determine whether dermal elements exist. If they are present, the intracranial portion is addressed.
- For lesions with an intracranial component, a follow-up MRI is recommended yearly for 3 years to rule out recurrent growth of any dermal remnants that were missed during the initial surgery.
- If a portion of the cartilaginous septum is removed surgically when following the sinus tract, it should be repaired during the same procedure to prevent a saddlenose deformity. If the defect is small, conchal grafts can suffice; if the defect is large, a costal graft may be required.

- If there is a large defect in the frontal bone, the occipital bone can be switched to the frontal position to give a smooth contour to the forehead. The frontal bone defect is repaired with allograft and placed in the occipital position.
- Nasal hemangiomas can be confused with venous malformations or arteriovenous malformations. If physical examination does not clarify the diagnosis, MRA with venous phase views can be helpful.
- If a patient has a history of nosebleeds without ulceration of the external component, a significant intranasal component may be present. An MRA of the nose and face can reveal the internal extent of the lesion.
- When nasal hemangiomas are treated with laser therapy, more treatments with less intensity decrease ulcerations and burns. Power settings should be increased incrementally over several treatments until the minimal effective power settings are determined.
- The Cyrano nasal tip deformity is a common sequela of hemangiomas of the nasal tip, with the involuting lesion leaving excess fibrofatty tissue with splayed alar cartilage domes.
- Costal grafts are notorious for warping, but steps can be taken to prevent this. All of the perichondrium is removed, and a 0.4 mm K-wire is inserted and cut just below the surface of the cartilage. Alternatively, the rib cartilage can be cut longitudinally into thin layers that can be “bruised” with a cartilage crusher. These cartilage sheets can be stacked and sutured together to achieve the desired dorsum height.
- Standard radiographs do not always accurately reveal nasal fractures in pediatric patients, because bones can undergo greenstick fracture rather than an outright fracture. In addition, the cartilaginous septum cannot be seen on plain radiographs. Accurate initial clinical assessment for significant bony and cartilaginous deviation and septal hematoma is crucial. If swelling prevents satisfactory assessment, the surgeon should ask the patient to return in 5 days with an old anteroposterior photograph to use for comparison.
- The septum can be splinted with sterile radiographic film cut to fit and sutured with the septum in the middle. This is removed after 7 days.
- If secondary surgical correction is necessary after initial closed reduction in adolescents, the surgeon should allow 9 to 12 months for complete healing before rhinoplasty. This interval can reveal areas of dorsal cartilage loss or deviation that may not be apparent initially.

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36

Primary Rhinoplasty in Adolescents: Indications and Techniques

Daniel R. Butz • Jeremy P. Warner



Adolescence is a critical time period for psychological and physical development. Nasal deformities can be a key source of distress, and many teenagers seek to undergo elective rhinoplasty.¹⁻³ Rhinoplasty is the most common elective cosmetic surgery performed in the adolescent population.⁴ Patients may present for aesthetic deformities, functional nasal airway obstruction, or both. Some people think that adolescents lack the intellectual maturity and insight to make decisions that will change their appearance forever.⁵ However, rhinoplasty has been found to be safe and effective and provides satisfactory patient outcomes.^{1,6}

INDICATIONS

In adolescent patients, surgeons must assess psychological maturity, desires, and expectations. This assessment should be coupled with the support and expectations of the parents. Patients should be near physical maturity to prevent disruption of nasal growth. Akgüner et al⁷ reported that females have adult nasal proportions and growth by 13 years of age and males by 15 years of age. In general, females should be 1 year postmenarche with stable growth.

Although the focus of rhinoplasty is generally on the aesthetic deformities and outcomes, patients with functional deformities often have significant airway obstruction. Those who present with specific and realistic requests are more likely to be satisfied with the postoperative outcome.⁸ Ideal patients have identifiable deformities, such as a dorsal hump, nasal deviation, a hooked tip, a pollybeak deformity, or a large nose. Surgeons should proceed cautiously with a patient who

brings in magazine clippings and requests to have a specific celebrity's nose.⁹ Similarly, those who present with minimal deformity but describe significant social, occupational, or functional disturbances should be questioned further and a psychological referral made when necessary. Body dysmorphic disorder typically develops during late adolescence.¹⁰ Finally, patients with an ethnic nose who seek a "white" nose should be counseled about realistic expectations.^{2,9}

PREOPERATIVE ASSESSMENT AND CONSULTATION

A preoperative assessment should include a thorough history and physical examination. A history of psychiatric illness is a relative contraindication to surgery, and unrealistic expectations are an absolute contraindication to surgery. Both the patient and parents must have similar expectations.

A thorough physical examination of the internal and external nose is performed.^{11,12} The skin type, thickness, and presence of any lesions or scars are noted. The dorsal aesthetic lines from the medial brows to the tip of the nose are inspected, and the overall shape and width of the tip are evaluated. From the lateral view, the radix position and dorsal profile are examined in the upper and middle thirds. Dorsal deformities, such as a dorsal hump, or saddlenose deformity, are noted. The tip length, projection, and rotation are analyzed. The infratip appearance, columellar position, nasolabial angle volume, and alar-nostril shape and size are also considered. A thorough functional examination will include an evaluation of the septum, inferior turbinates, internal nasal valves, and external nasal valves.

Surgeons must take photographs at the initial consultation and review them with the patient. This allows patients to express their concerns in detail and to clarify the surgical goals.¹³ Standard photographs include frontal, oblique, lateral, and worm's-eye views of the face. Using modern computer software, surgeons can show patients how their nose might look after the surgery is complete.^{14,15} Although computer software can be a powerful tool for communication between a surgeon and patient, realistic expectations and limitations must be thoroughly discussed. I (J.P.W.) use three-dimensional imaging in my office to work toward a common goal with the patient. I stress that healing can cause the actual final outcome to vary from the final outcome shown on the computer, and that the computer image is not meant to show promised final results.

High patient satisfaction and low revision rates are obtained when surgeons spend quality time during the initial consultation discussing the patient's concerns and goals, along with their own expectations and limitations. This is a critical time in the physician-patient relationship; the surgeons must ensure that they understand their patients' desires and that patients understand what can and cannot be achieved. If the patient and surgeon are "on the same page" at the end of the consultation process and the surgeon thinks that he or she can provide the results that have been discussed, the likelihood of an optimal outcome is very high, and surgery can proceed.

ANESTHESIA AND PREOPERATIVE MANAGEMENT

Most patients undergoing rhinoplasty are treated on an outpatient basis with general anesthesia. Perioperative antibiotics are routinely given, but postoperative antibiotics are not necessary. The internal and external nose is infiltrated with local anesthesia consisting of a 50:50 mix of 1% lidocaine with 1:100,000 epinephrine and 0.25% bupivacaine with 1:200,000 epinephrine. Usually 2 to 5 ml is used and provides adequate pain control and vasoconstriction without causing excessive tissue distortion. Oxymetazoline-soaked (Afrin) pledgets are also packed into the nares to provide additional mucosal vasoconstriction. Patients typically receive 6 to 10 mg of intravenous dexamethasone intraoperatively, followed by a 6-day oral tapering dose of methylprednisone. The face is prepared with a gentle cleanser such as Cetaphil.

SURGICAL APPROACHES

Incisions and Approaches

Every surgeon has different comfort levels with open and closed approaches. In this chapter we will focus on the open approach, because it typically provides more exposure and control over the final outcome. Several different incisions are used for open rhinoplasties. We have found that a gull-wing incision heals better than a stairstep or straight transverse incision (Fig. 36-1). It should be placed at the midcolumnellar level above the medial crural feet and meet the vertical marginal incision at a right angle. A converse scissors is then used to elevate the soft tissue flap off of the medial crura (Fig. 36-2, *A*).

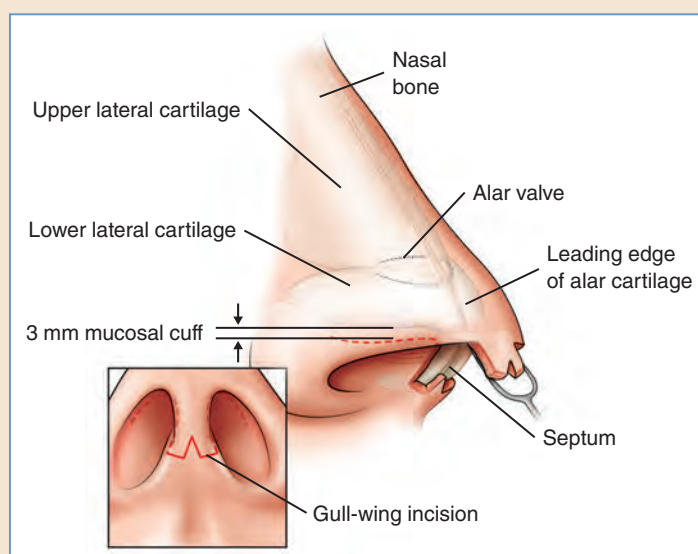


Fig. 36-1 Nasal anatomy and gull-wing incision.

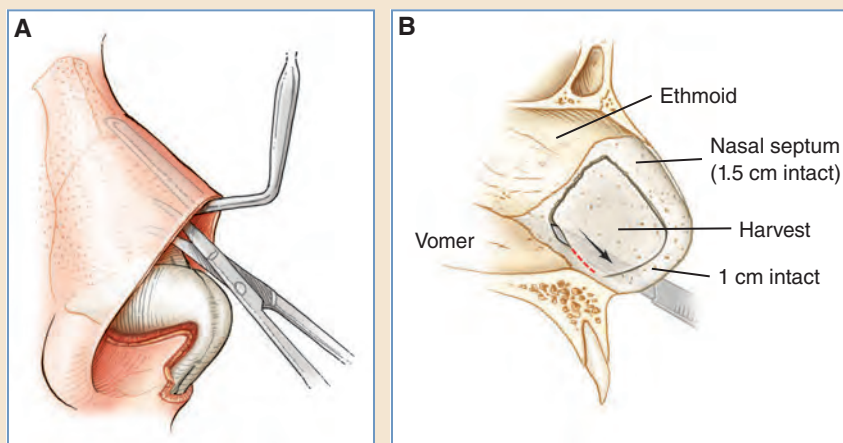


Fig. 36-2 Flap elevation and harvesting of septal cartilage. **A**, A skin envelope is raised with uniform thickness. **B**, The central septum is harvested with a No. 15 blade. A 1 cm L-strut is preserved to prevent a saddlenose deformity and preserve tip support.

Septal Deformities and Cartilage Harvest

Septoplasty is typically performed first to fix any functional deformity and/or to harvest cartilage if necessary. If tip work is planned, the septum can be harvested through an open approach. However, if patients are satisfied with their tip characteristics, the cartilage can be harvested through a hemitransfixion incision to prevent disruption of the nasal tip supporting structures (which may introduce unwanted variability in tip aesthetics).

The anterior septal angle is easily identified through an open approach. The soft tissue between the medial crura can be removed to reduce fullness in the nasolabial angle. After the anterior septal angle is exposed, a No. 15 blade is used to make a small incision through the perichondrium on the septal cartilage to access the submucoperichondrial layer and allow easy flap elevation with a Freer elevator in an avascular plane. After both mucoperichondrial flaps have been adequately elevated, the septum can be harvested. A No. 15 blade is used to excise the central portion of the cartilage, leaving behind an L-strut that is at least 1 cm in width (see Fig. 36-2, *B*). The surgeon must take into consideration any dorsal hump resection that may be planned, because this will further reduce the dorsal L-strut. The wide exposure gives adequate access to remove any septal bony spurs, because these can cause significant airway obstruction.

Swinging-Door Flap

Patients with caudal septal deviation resulting from caudal septal base deflection off the maxilla frequently require a swinging-door flap.¹⁶ The caudal septal strut can be released from the maxilla, which will allow the inferior caudal septum to swing into the midline and rest in the maxillary groove. This may require excising 1 to 2 mm off the base to allow it to freely move to the midline without residual curvature. A caudal septal cartilage strut graft will provide additional support and structure to the caudal septum if necessary.

Septal Straightening Techniques

A crooked dorsal septum can create asymmetries of the brow-tip aesthetic line. These can be particularly difficult to correct. Several techniques can be used to correct or camouflage these deformities. Gentle scoring of the septal cartilage on the concave side will cause the cartilage to bend away from the scoring. Castellation or full-thickness incisions that weaken the septum will often disrupt the intrinsic bending forces (although if used should be reinforced with additional strut grafts to support the septum and hold it straight). Spreader grafts can also be employed to support a crooked septum and provide additional structural support. Often, a simple spreader graft is insufficient, and complete dorsal septal reconstruction is necessary. This may require complete excision of the native septum and dorsal septal graft reconstruction.¹⁷ Finally, onlay grafts can also be used to camouflage the defect.¹⁸

Nasal Tip Reconstruction

After the septal work has been completed, the major tip parameters should be set: length, projection, and rotation. I (J.P.W.) prefer to set these parameters first, and then shape the dorsum to match the ideal tip position already created. The tripod concept is applied when setting these parameters^{19,20} (Fig. 36-3). The tripod concept treats the lower lateral cartilage as a tripod, with each lateral crura representing one leg of the tripod, and the medial crura is the third leg. Shortening or lengthening a leg will affect the location of the tip. After these parameters are set, it is much easier to shape the dorsum in relation to the tip. Fine-tip sutures and additional grafting can be performed last to prevent disruption during dorsal manipulation. This approach provides predictable results.²¹

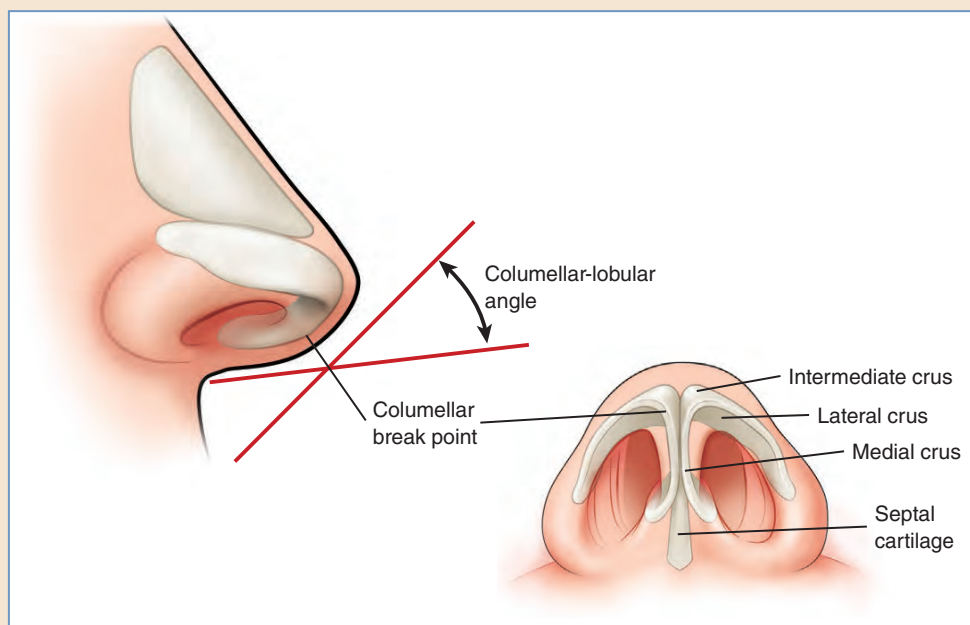


Fig. 36-3 Lower medial and lateral cartilage anatomy. The tripod theory treats each lateral crus and the paired medial crura as the legs of a tripod. Manipulation of each “leg” will affect tip position and projection.

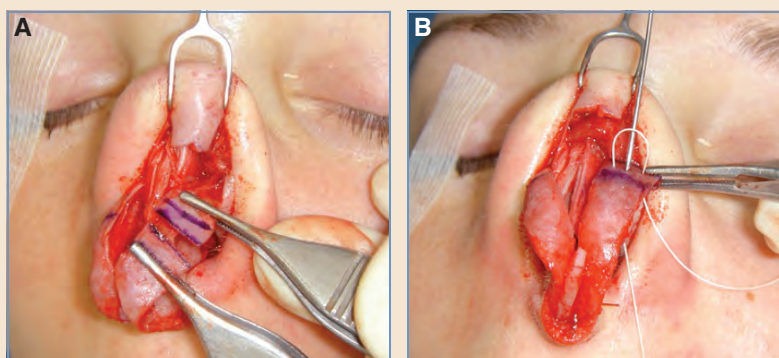


Fig. 36-4 Lateral crural flap. **A**, The lateral crura have been released from the underlying mucosa and transected. **B**, The lateral crura are sutured together in an overlapped fashion with a Vicryl horizontal mattress suture. The flap is used to rotate the tip up and deproject the nose.

Nasal Length

Nasal length is determined by the distance from the nasofrontal angle to the tip-defining point. The nose can be shortened with two techniques: rotating the tip up and deepening the nasofrontal angle. Nasolabial augmentation can also make the nose appear shorter by increasing the apparent rotation. The tip can be rotated by shortening the lateral crura with the lateral crural flap technique (Fig. 36-4). The flap is usually only 2 to 3 mm in length but can vary depending on how much rotation is needed. The amount of flap overlap is measured bilaterally with calipers.



Fig. 36-5 Medial crural flap. **A**, The medial crura are measured and incised. **B**, Both medial crura have been overlapped and sutured together. **C**, This flap results in derotation and deprojection of the tip.

The lateral crura are cut near the middle and then overlapped, shortening the lateral crura. The cartilage flaps are sutured together with 5-0 Vicryl horizontal mattress sutures.

Short noses frequently have dorsal deficiency and require augmentation. Derotation, infratip lobule grafts, and nasofrontal augmentation will increase the length of the nose. Surgeons can also increase the apparent length of the nose by using dorsal onlay grafts and by deepening the nasolabial angle. Bilateral extended spreader grafts and caudal septal extension grafts may also be necessary to significantly lengthen a short nose.

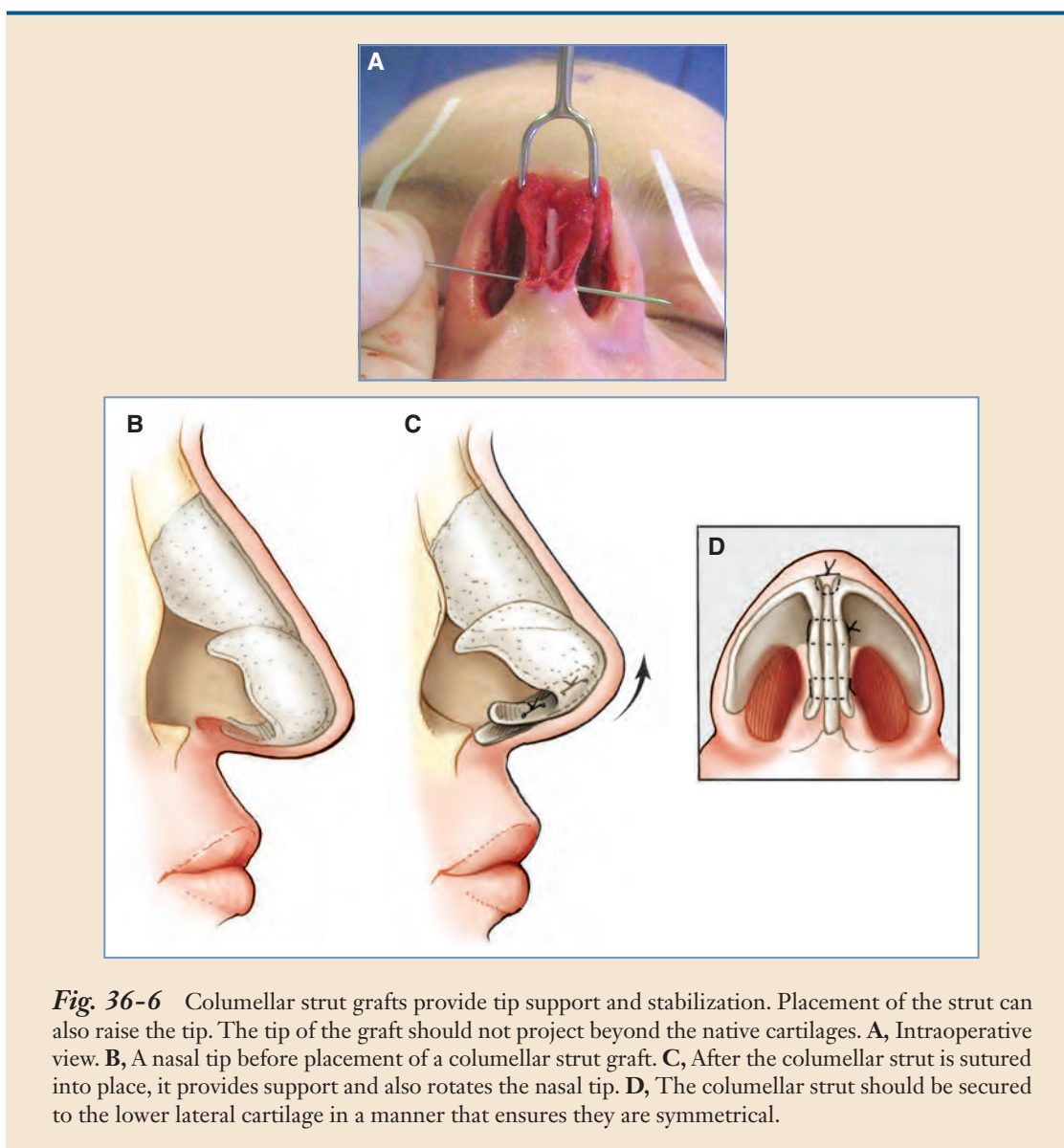
Projection

Overprojection is one of the more difficult parameters to correct and is frequently neglected, leading to dissatisfaction. If the medial or lateral crura are too long, they should be shortened with lateral crural flaps and/or medial crural flaps. A medial crural flap procedure is similar to a lateral crural flap procedure, but the medial crura are incised near their cephalic one third (Fig. 36-5). Deprojection of the nasal tip can create alar base widening and alar flaring, which should be planned for and evaluated during surgery to determine whether soft tissue alar base reduction will be necessary.²² Potential soft tissue excision techniques of the alar base should be discussed with patients preoperatively, because additional external sutures and a scar will be present (although these scars heal incredibly well when the procedures are performed properly).

Underprojection is frequently a problem in nonwhite noses, particularly in blacks and peoples of Asian and Hispanic descent. Projection can be increased by columellar strut placement, transdomal suturing, lateral crural steal, and tip grafting.^{12,23,24}

Rotation

Tip-rotation issues can cause several different problems. An underrotated tip gives the appearance of a long nose. This can be caused by short or weak medial crura creating ptosis. A columellar strut can be sutured between the medial crura to provide support and rotate the tip up. It should extend down to the nasal spine to provide strength and support to the tip (Fig. 36-6). This can be secured by placing a horizontal mattress suture of 4-0 Vicryl on a Keith needle. The first pass should include both medial crura and the columellar strut graft, and the second pass goes behind



the columellar strut. As seen in Fig. 36-6, it is important to retract the nose with a double-prong skin hook to ensure that equal tension is placed on each dome.

An overrotated tip often creates excessive nostril show in the AP view, giving a “pig nose” appearance. This deformity can be corrected by several methods: an infratip lobule shield-type graft increases nasal length, shortening the medial crura and excising soft tissue at the nasolabial angle. Deepening the nasolabial angle in effect decreases the nasolabial angle, creating the appearance of a longer nose, and the tip rotation. In severe cases the supporting attachments from the tip and upper lateral cartilages and septum will need to be released and the tip rotated downward and secured in place with extended spreader grafts, thus forcing the tip down and keeping it in place.

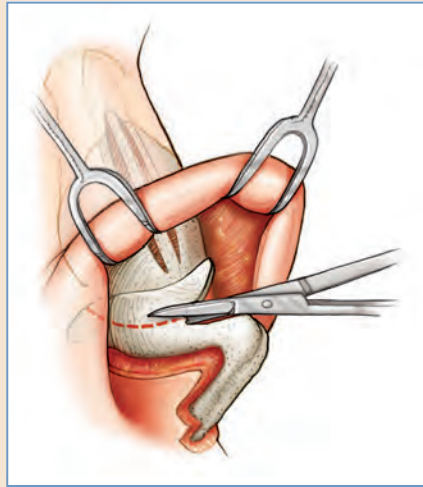


Fig. 36-7 The lower lateral cartilages are trimmed along the cephalic margin, preserving the mucosa and 6 to 7 mm of rim for external valve support. The surgeon should preserve the upper lateral cartilage attachments to the septum at the nasal valve.

Cephalic Trim

Wide lateral crura can contribute to a widened bulbous appearance of the tip and supratip fullness. These deformities can be improved by a resection of the cephalic margin of each lower lateral cartilage (Fig. 36-7). The resection must be symmetrical and leave at least 6 to 7 mm of lateral crura to support the external nares and prevent external valve collapse.²⁵

DORSAL MANIPULATION

Dorsal Hump Reduction

Dorsal hump deformities are generally corrected after the major tip parameters are set. The dorsum can be shaped to match the tip projection and rotation (Fig. 36-8). The nasal dorsum is composed of the bony upper nasal vault and the cartilaginous middle nasal vault. When a nasal hump is present, the bony upper nasal vault is rasped to achieve a straight profile (Fig. 36-9). This should be done in a controlled, stepwise fashion, assessing progress throughout the procedure. The surgeon must not overresect the profile and create an unnatural appearance. If a component of cartilaginous dorsal excess at the middle nasal vault is also present, a sharp straight scissors or No. 11 blade can be used to achieve an ideal profile in this area. Prominent residual dorsal septal cartilage will result in a pollybeak deformity. A rasp will not remove the cartilage profile. Proper profile assessment requires the surgeon to squeeze out the edema with a cold wet sponge and sit on a stool to have a perpendicular view of the dorsal profile. Typically, altering the dorsal profile to remove a prominent hump will result in an open-roof deformity, which must be corrected with osteotomies (see Nasal Bone Osteotomies).



Fig. 36-8 A-C, Preoperatively this female patient thought that her nose was too big for her face, and she was unhappy with the nasal hump on profile view. She had nasal airway obstruction. D and E, Primary open septorhinoplasty was performed. The surgical steps included a septoplasty, excision of soft tissue at the nasolabial angle, tip deprojection with vertical lobular division (medial crural overlay), a columellar strut for tip stabilization, a bilateral cephalic trim, tip sutures, hump takedown, medial and lateral osteotomies, and bilateral spreader graft placement. F-H, The patient is shown 1 year postoperatively.

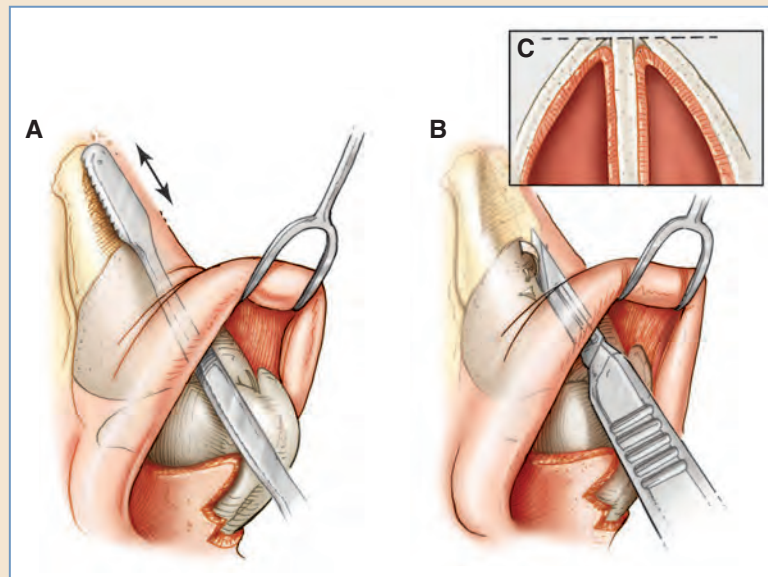


Fig. 36-9 Dorsal hump reduction. **A**, A rasp can be used for the bony component. **B**, A scalpel or straight scissors is used for the cartilaginous dorsal resection. **C**, It is essential to leave a smooth contour between the upper lateral cartilage and the septum after hump takedown is completed.

Dorsal Augmentation

Augmentation of the dorsum may be necessary to set a proper nasal height, correct a saddlenose deformity, and improve the dorsal nasal contour. If the dorsal height is too low, it can result in an overresected appearance. Autologous materials are preferred for dorsal augmentation. Septal cartilage can provide a strong, stable graft, but a large piece of cartilage or stacked cartilage grafts is often needed. Costal cartilage is frequently used.²⁶ The graft should be carefully sculpted to fit the defect, and the edges must be beveled to prevent visible step-off deformities and palpability. The graft must be secured to prevent malposition. We prefer to secure the graft to the nasal skin as described in the Onlay Grafts section. Saddlenose deformities are rarely encountered in patients who present for primary aesthetic surgery and are typically the result of trauma, previous surgery, or a congenital deformity.²⁷

Spreader Grafts

Spreader grafts are frequently used to increase the internal nasal valve angle, strengthen the dorsum if the L-strut is fractured, and straighten the dorsal aesthetic lines. These grafts are placed between the septum and upper lateral cartilage (Fig. 36-10). They should be sutured in place with

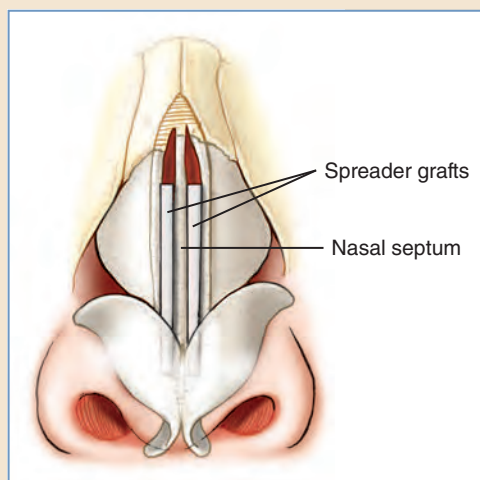


Fig. 36-10 Spreader grafts are useful to obtain optimal dorsal aesthetic lines and to open the internal nasal valve.

4-0 Vicryl horizontal sutures spanning the upper lateral cartilage, spreader graft, and the septum. At least two sutures are placed to prevent the graft from rotating out of position.

Nasal Bone Osteotomies

Nasal bone anatomy can be quite variable. Younger patients have much thicker bone compared with elderly patients. In patients with a history of trauma, the bone may be thick and callused. There are three indications to perform osteotomies: to straighten crooked nasal bones, to close an open-roof deformity that is frequently created from the dorsal hump reduction, and to narrow a wide upper third^{28,29} (Fig. 36-11).

Medial osteotomies are made first with a fading angle laterally to prevent a rocker deformity (seen when the medial osteotomies are carried straight up the bones). An open technique provides optimal exposure to visualize the placement of the osteotome. Additional local anesthetic is infiltrated in the internal nasal mucosa at the piriform aperture just superior to the head of the inferior turbinate in preparation for lateral osteotomies. A No. 15 blade is used to make a stab incision at this site. A Freer elevator can be used to create a subperiosteal tunnel for the osteotome, which helps to decrease bleeding. Curved, guarded osteotomes are used to make the lateral osteotomies in a high-low-high fashion, beginning superior to the inferior turbinate at the piriform aperture. If it is placed lower, it will cause the inferior turbinates to drop into the airway and obstruct the airway. The osteotomy is made in a curved fashion, aiming and ending medial to the medial canthus but not meeting all the way to the medial osteotomy (Fig. 36-12).

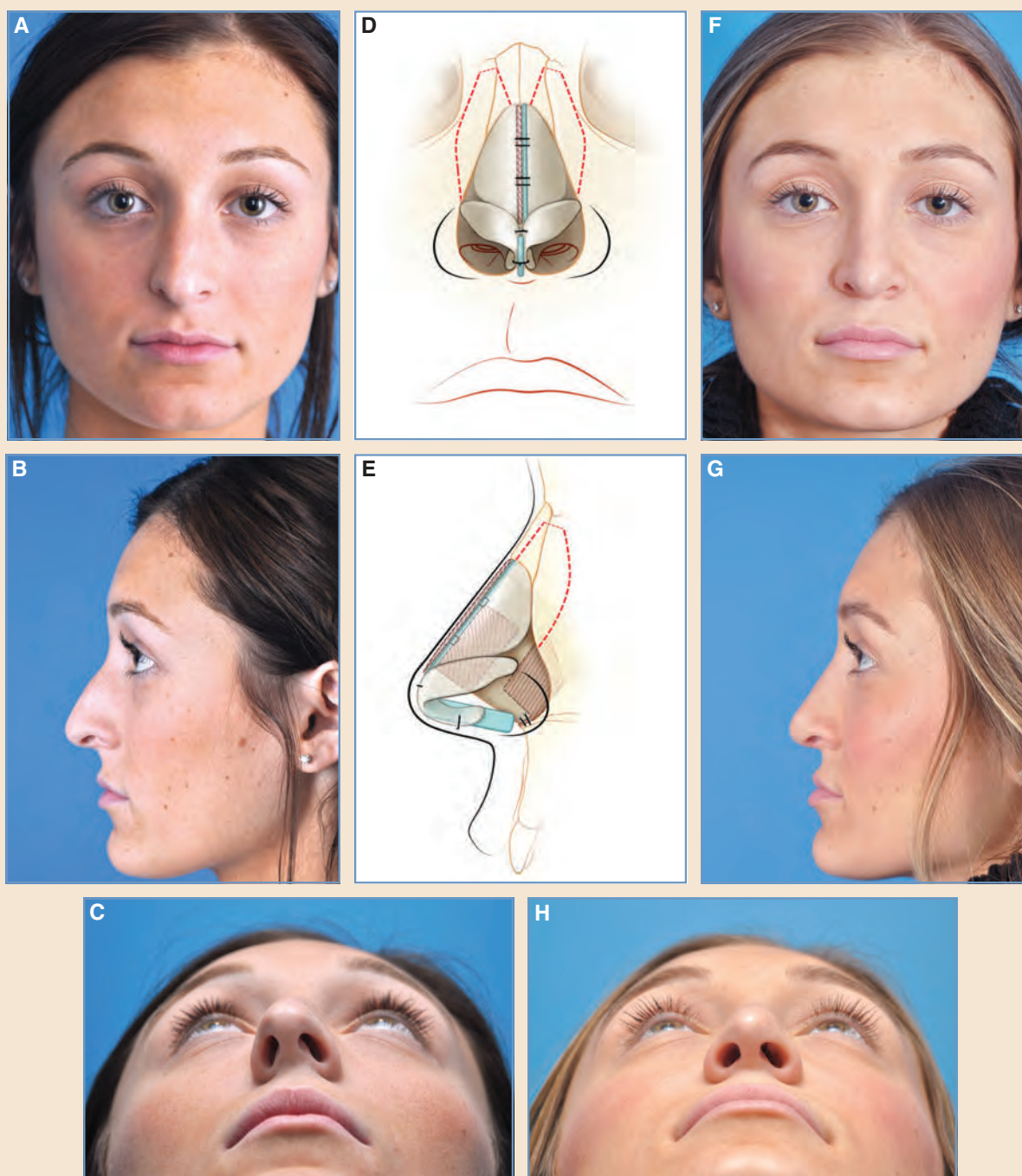


Fig. 36-11 A-C, Preoperatively this female patient's primary concerns were nasal obstruction and the appearance of the dorsal hump on profile view, and she thought that her nose was crooked. She did not want any major changes to her nasal tip. D and E, Primary open septorhinoplasty was performed. Surgical steps included septoplasty with submucous resection of cartilage and bone, dorsal septal reconstruction with a dorsal replacement graft, medial and lateral osteotomies, dorsal hump takedown, and a columellar strut to straighten and maintain the tip dynamics. F-H, The patient is shown 1 year postoperatively.

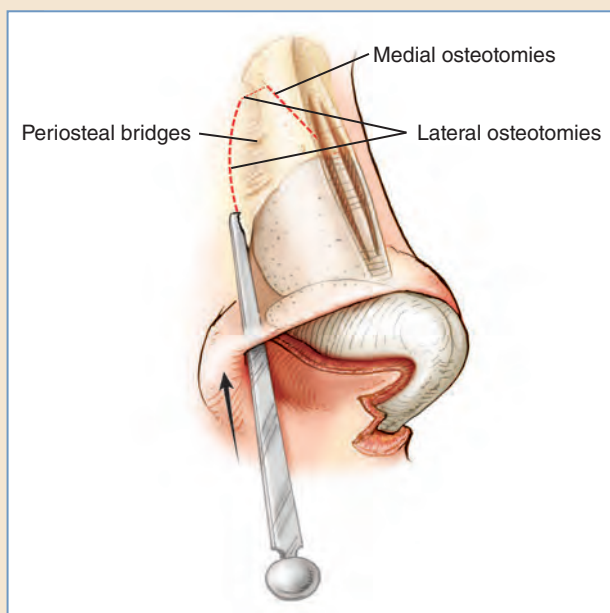


Fig. 36-12 Nasal bone osteotomies. Medial osteotomies are performed first; the surgeon is careful to not disrupt the septal nasal bone junction. Lateral osteotomies are started superior to the inferior turbinate and performed in a high-low-high fashion. The osteotome can be treated as a lever to assist in creating a greenstick fracture of the nasal bone medially.

On completion, the osteotome is used as a lever to create a greenstick fracture between the medial and lateral osteotomies, and the nasal bone is brought in medially in a controlled and stable fashion. If the nasal bones need to be widened, the same maneuvers are performed, and the bone is then lateralized and supported intranasally with a small piece of Surgicel.

NASAL FINE-TIP WORK

The nasal fine-tip work should be done toward the end of the procedure. This will decrease the chance of disrupting fine suture and graft placement during dorsal maneuvers.

Tip Sutures

A single-dome unit-binding suture (intradomal) can be used to create individual refinements to each dome, set the exact tip position and angle, and create more definition.¹² The use of this suture can ensure that both tips have a similar anatomic shape. A double-dome unit-binding suture (interdomal) can be placed between the domes to medialize the domes, narrow the tip, and bring the domes together to a symmetrical height.²³ These maneuvers can also increase the tip projection. Another commonly used tip suture is an intermediate crural horizontal mattress suture. This suture can strengthen the medial crural–columellar strut complex and narrow the infratip columella to decrease a hanging infratip. All of these techniques should be done in a stepwise fashion, continuously assessing their impact on the overall nasal appearance.

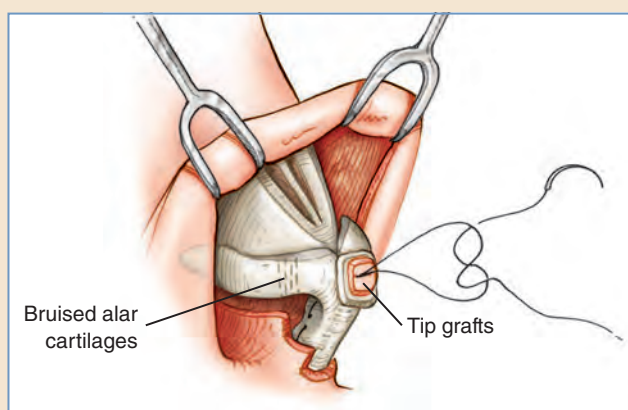


Fig. 36-13 Nasal tip shield grafts are placed to add length or rotation when needed. Tip grafts are best secured to the nasal cartilaginous skeleton.

Tip Grafts

Tip grafts can be used to camouflage asymmetries, increase nasal projection, and increase or decrease tip rotation, depending on the location. Grafts need to be meticulously shaped to fit the desired defect and to effect an exact change. The edges must be beveled to ensure that there are no palpable step-offs that may become visible when the swelling decreases. Most tip grafts are best secured directly to the nasal skeleton with 6-0 sutures.

An infratip graft placed just below the domal angle will add fullness and definition to the infratip region (Fig. 36-13) and result in an apparent derotation of the tip. A cap graft can be placed spanning both domes to add projection or definition. They can also be placed over a single dome to provide better domal symmetry.

Lateral Crural Strut Grafts

Lateral crural strut grafts are commonly used in patients who present with external valve collapse or misshaped native cartilages resulting in deformity. These grafts are optimally created from strong septal or rib cartilage to provide the necessary strength. They are typically placed under the native lower lateral cartilage (between the cartilage and mucosa) but can also be placed in an onlay fashion. The medial aspect should not extend past the domal apex, and a smooth transition laterally is essential to prevent graft visibility after swelling subsides.³⁰

Onlay Grafts

Before final closure, the nasal sidewalls and dorsum are assessed for any irregularities or asymmetries that could lead to a suboptimal outcome. Deformities of this type can be corrected by placing a dorsal onlay graft. This is done after all of the maneuvers have been completed, because very fine grafts will usually need to be created. Unlike tip grafts that are sutured to the nasal skeleton, dorsal onlay grafts can usually be more precisely secured with a percutaneous horizontal mattress suture through the skin and graft. The suture should be tied securely but should not indent the skin to prevent ischemia as postoperative swelling develops (Fig. 36-14).



Fig. 36-14 A-C, Preoperatively this female patient thought that her nose was too big for her face. She had nasal airway obstruction. D and E, She had a primary open septorhinoplasty, which included septoplasty (with submucous resection and caudal base repositioning), dorsal septal reconstruction with a replacement graft on the left side, right spreader graft placement, tip deprojection with vertical lobular division (medial crural overlay), stabilization of the tip with a columellar strut, tip sutures placement, excision of soft tissue at the nasolabial angle, cephalic trim, dorsal hump takedown, medial and lateral osteotomies, and dorsal sidewall onlay grafting. F-H, She is shown 1 year postoperatively.

FINAL REFINEMENTS: EXCISION SOFT TISSUE TIP TECHNIQUES

Excessive width of the lower third of the nose may be apparent on preoperative evaluation or may become obvious only when a tip is changed during surgery. Surgeons should point out these potential issues to patients and offer solutions that can create a well-balanced and aesthetically pleasing result.

Alar Base Surgery

The alar base has two components: a cutaneous ala portion and a vestibular surface. These can be managed independently to cause varying degrees of changes to the alar width and nostril size²² (Fig. 36-15). The optimal position for the incision is along the natural curve of the nasal sill to prevent unsightly scars. The nasal margin excision will determine the effect on the nostril and alar base. An external alar base reduction with an inverted-V resection can be used to reduce the columellar-alar base distance and the apparent nasal width. An internal alar base reduction with a V-shaped resection will narrow the nostril circumference but maintain the nasal width. Alternatively, to decrease the base width and nostril size, a square-shaped piece should be resected from the nasal margin.

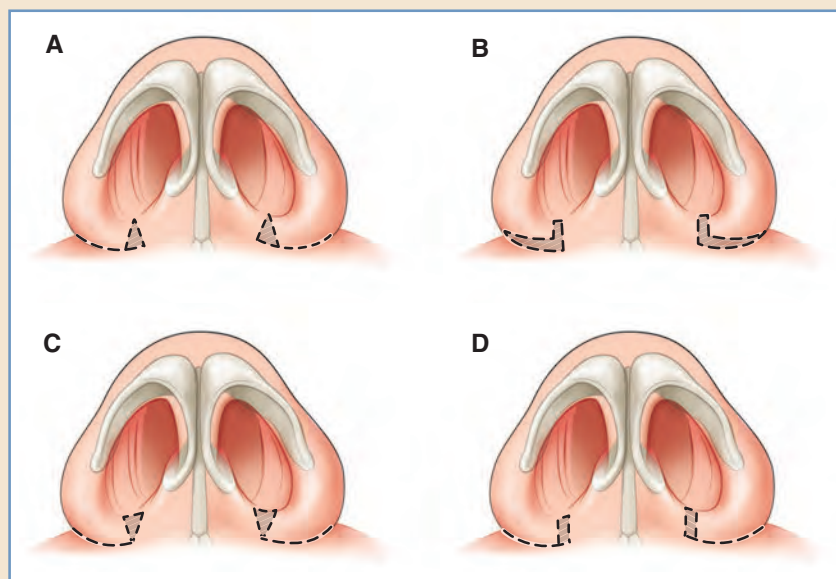


Fig. 36-15 Soft tissue resections of the alar base. **A**, External alar base excision. The excision is planned as an inverted-V to not disturb the circumference of the nostril margin. **B**, Internal base excision. Isolated excision of the nostril margin reduces only the nostril circumference while leaving the columellar-alar base distance unchanged (thus preserving the alar base width). **C**, External and internal base excision. **D**, Alar flare reduction. A segment of tissue is removed from around the alar base to reduce the tip-alar base distance, thus shortening that limb and making the ala straighter.

Alar Flare Reduction

After the alar base width and nostril size have been addressed, the flaps are mobilized and advanced medially in position with one stitch. Alar flaring can then be assessed and managed. This is best done in the basal view. Ideally, the alae should be relatively straight from the nasal tip to the base. If they are excessively flared or convex, excising a portion of the alar base and shortening the distance can straighten them.²² These incisions are closed with 6-0 nylon interrupted sutures.

Alar Hooding Reduction

Alar hooding is seen when the alae hang excessive low and cover the columella in the profile view. In these patients, an idealized alar curvature is drawn, and an elliptical excision is made along the border of the nostril margin. The incision should be along the visual border, because these tend to heal well.²² The incision is closed with 6-0 running nylon sutures.

CLOSURE

The columellar incision is closed with simple 6-0 nylon sutures. The junction at the transverse columellar and vertical marginal incisions is at the most risk for poor scarring resulting from a trap-door effect. Minor undermining of the columellar skin on the lateral aspect of the columella and placement of a partial-thickness suture superiorly and a full-thickness suture inferiorly to tuck in the superior flap edge decrease the risk of the trap door effect. The marginal incisions are closed with 4-0 chromic suture. Care is needed to not distort the alar rim, because this can create unfavorable scarring.

Silicone septal splints are placed to stabilize the septum and secured with a 4-0 Vicryl transfixion suture. Nasal packing is unnecessary, because it typically is extremely uncomfortable for the patient and provides no benefit. If osteotomies are performed, an Aquaplast cast is placed to stabilize the nasal bones.

POSTOPERATIVE CARE

Patients are provided with narcotic pain medications and instructed not to start NSAIDs or aspirin products until 48 hours after surgery. Oral steroids are given in a 6-day tapered dose. Oral *Arnica montana* is also used to decrease bruising. Patients are encouraged to keep their heads elevated and to aggressively ice the area. They are instructed not to engage in physical activity for 3 weeks and contact sports for 6 weeks.

Sutures are removed from the columella 4 to 5 days postoperatively, and small Steri-Strips are placed and removed after 4 days. The cast and splints are removed 7 to 8 days after surgery. Patient follow-up is at 3 weeks, 3 months, 6 months, and 1 year. Standardized photographs are taken at each follow-up appointment.

MANAGEMENT OF COMPLICATIONS

Careful preoperative planning and surgical technique with conservation of the tissue can minimize postoperative complications. In general, the complication rate from rhinoplasty surgery can be quite low, but complications will arise. Surgeons must be supportive and responsive to patients' concerns.

Complications range from postoperative hematomas and infection to aesthetic deformities that are frequently more difficult to treat and may require revision operations.³¹ Both moderate swelling and periorbital ecchymosis are expected but can be minimized with constant icing, *A. montana*, and steroids. Mild postoperative oozing is expected and can be controlled with a mustache dressing for 24 to 48 hours. More moderate bleeding can be managed with head elevation and ice on the back of the neck. More serious bleeding may require reexploration in the operating room. Unrecognized septal hematomas can lead to cartilage necrosis and loss of support. The risk of septal hematoma can be decreased by placing horizontal mattress sutures in the septum to reduce dead space and by creating a small, unilateral stab incision in one (not both) of the mucoperichondrial flaps to allow blood to drain.

Infections are rare but can range from mild cellulitis to abscess formation.^{31,32} Early identification and treatment are essential to prevent tissue necrosis and potential toxic shock syndrome.³³ Mild cellulitis will normally respond to cephalosporins. If the cellulitis persists or worsens, admission for IV antibiotics may be appropriate.

Postoperative deformities may commonly result from underresection or overresection of the osteocartilaginous framework, unrecognized asymmetry at the time of surgery, postoperative bone movement, cartilage resorption, scar tissue formations, cartilage warping, and poorly shaped grafts with edges or graft migration resulting from securing issues.³¹ A suboptimal outcome can occur for other reasons. Typically, major deformities recognized early do not improve over time. However, frequently, with more minor deformities, what appears to be a deformity may actually be the result of swelling, and patient waiting is always prudent. Patients can attempt to perform molding techniques to manually shape the cartilage by applying controlled pressure on the defect for 5 minutes, five times a day, for 5 weeks. Taping and massage may also help. Revision operations can be considered for persistent defects present for more than 1 year. For a patient who is concerned about a persistent deformity after 1 year, the surgeon should correct the deformity if he or she thinks this is possible.

CONCLUSION

Adolescence is a critical period of both physical and mental development, and self-image can be adversely affected by nasal deformities. Adolescent rhinoplasty can result in gratifying outcomes that greatly enhance a patient's psychosocial well-being. In many patients, functional and aesthetic improvement can be achieved. A thorough preoperative evaluation and discussion of expectations, coupled with computerized imaging, can prevent misunderstandings that lead to postoperative disappointment. A stepwise approach to nasal surgery is important, with an emphasis on a conservative but thorough approach to each patient's aesthetic and functional needs. By following these steps, surgeons can fully address their patients' concerns in a logical and systematic fashion to obtain the best possible outcome.

KEY POINTS

- The nose is the focal point of facial aesthetics; therefore it is a key component in establishing harmonious facial balance.
- An adolescent's self-esteem is greatly influenced by physical appearance.
- For all patients undergoing rhinoplasty, especially adolescents, communication between the patient, parents, and plastic surgeon is essential for a successful result. Reasonable expectations must be discussed preoperatively with the patient and family.
- A stepwise approach to rhinoplasty is recommended, allowing a sequential surgical treatment plan that is predictable and reproducible.
- Surgeons should define the tip parameters (length, projection, and rotation) and then match the dorsum to the tip.

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37

Pediatric Head and Neck Masses

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Pediatric head and neck masses are a heterogeneous group of lesions of various etiologies. Diagnosis of these lesions can be challenging for several reasons. First, not all lesions are readily apparent, and children may be unable to adequately communicate their symptoms. Second, with such a diverse group of pathologies, practitioners may not have first-hand experience with many of these diagnoses. Finally, the anatomy of the head and neck region is a complex, dense network of vital structures. In children the size and proportion make physical examination more difficult.

Head and neck masses in children may be classified by their anatomic location, age at presentation, and cause. In this chapter, we will categorize lesions as congenital, infectious, inflammatory, and neoplastic.

PATIENT HISTORY, PHYSICAL EXAMINATION, AND IMAGING

To diagnose pediatric head and neck lesions, we begin with a thorough history and physical examination. Obtaining a history involves getting patients' and/or their caregivers' account of symptoms. Information gleaned should include onset of mass appearance, location, growth, associated local and constitutional symptoms, recent illness, sick contacts, recent travel, contact with animals, trauma, vaccination record, drug use, a personal or family history of cancer, and head and neck pathology.

The timing of the onset of symptoms may help to identify a congenital lesion and narrow a patient's differential diagnosis. Surgeons should keep in mind, however, that many congenital lesions present at birth are not noticed until later in infancy or childhood. Temporal relationships and changes over time are also important with acquired lesions. Some infectious and inflammatory lesions such as cervical lymphadenopathy may be preceded by a recent upper respiratory tract illness or other recent exposure. The rate of growth of neoplasms alerts practitioners to aggressiveness of the tumor. The location and laterality provide further clues for diagnosis.

In a physical examination, the appearance of a lesion at rest and with diagnostic maneuvers such as swallowing is noted. Associated skin changes such as erythema and overlying punctum are observed. On palpation, lesion texture and depth and adherence to underlying structures or skin may be appreciated.

Imaging is an important adjunct to a history and physical examination to help determine a diagnosis and to better understand the anatomic relationship for preoperative surgical planning. For most neck lesions and many head lesions, ultrasonography may be used as a first-line imaging modality. The advantages of using ultrasonography are that it is noninvasive, does not typically require sedation, delivers no radiation, and is able to distinguish cystic from solid lesions. The disadvantages include variability in results based on the ultrasonographer's skill and the inability to provide detailed soft tissue differentiation. CT and MRI are also frequently used. CT is particularly good at demonstrating bony pathology, whereas MRI provides superior soft tissue definition.

Treatment varies by the type of lesion. In general, congenital lesions may be managed surgically, and infectious and inflammatory lesions are treated medically. Treatment for neoplastic lesions is mixed. Some treatment protocols include chemotherapy and radiotherapy. In those cases, surgeons may still be called on to obtain tissue from an open tissue biopsy for diagnostic or staging purposes. Many treatment protocols include surgical tumor resection with or without subsequent adjuvant therapy.

CONGENITAL LESIONS

Congenital lesions make up a significant proportion of pediatric head and neck masses. In some series they are the most common cause of these lesions.¹ Despite their presence at birth, many of these lesions go undiagnosed until they become acutely inflamed, often later in infancy or childhood. With the improvement in imaging technology, congenital head and neck lesions are increasingly diagnosed in the prenatal period on routine ultrasonography. Prenatal MRI may be used to further characterize these masses. In addition to preparing parents for an eventual diagnosis, this information may be used to plan early intervention that can be lifesaving. If a head and neck tumor is diagnosed prenatally and considered potentially airway compromising, surgeons may perform an ex utero intrapartum treatment (EXIT) procedure. In an EXIT procedure, a modified cesarean delivery is performed. The baby is partially delivered and may undergo various procedures before separation from the umbilical cord and placenta. This allows continued oxygenation through the maternal circulation. In infants with head and neck masses, this is most frequently used as a means to secure the airway in a controlled fashion rather than emergently after delivery.

Vascular Malformations

Vascular malformations are lesions involving the vascular system that are present at birth and grow in proportion to the infant. They may or may not be visible at birth or in infancy and may remain quiescent or gradually enlarge. They are typically made up of mature vessels and can be classified based on vessel type and flow characteristics. They need to be distinguished from hemangiomas, which more commonly appear after birth and grow out of proportion to overall growth. Vascular

malformations are discussed in Chapters 11 and 13; however, because they are frequently part of the differential diagnosis of head and neck lesions, we also discuss them here.

Lymphatic malformations are highly variable, slow-flow vascular malformations that are characterized by disorganized, dilated lymphatic channels. The high concentration of lymphatics in the head and neck region explains why 75% of these malformations occur in this region. Previously known as *cystic hygroma* and *lymphangioma*, these are the most common vascular lesion affecting the head and neck region.² They may be classified as macrocystic (≥ 2 cm spaces), microcystic, or mixed (Fig. 37-1). Clinical staging is based on anatomic location³ (Box 37-1).

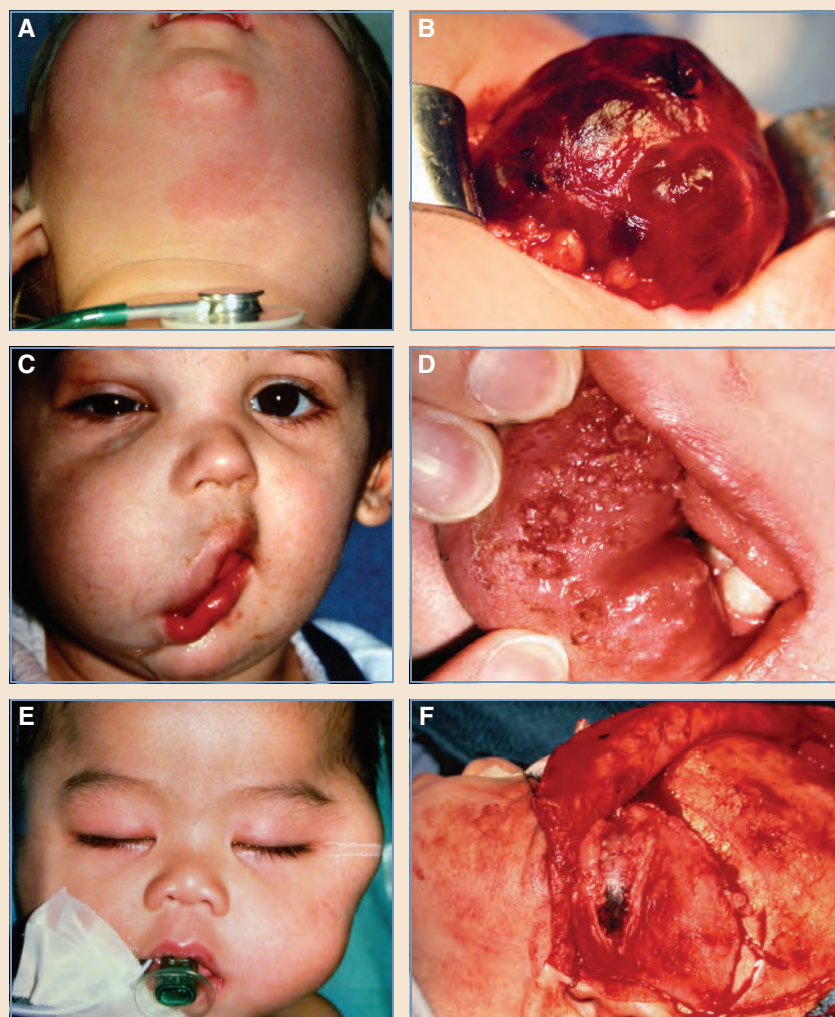


Fig. 37-1 A and B, A macrocystic lymphatic malformation seen in preoperative and intraoperative views. Although sclerotherapy may be the treatment of choice, surgery may be indicated. The large cysts of the malformation are readily visible here. C and D, A microcystic lymphatic malformation of the cheek, which is most easily diagnosed by the presence of fine lymphatic vesicles on the labiobuccal surface. E, A macrocystic lymphatic malformation replacing the entire right temporalis muscle and presenting with acute bleeding into the malformation. F, At resection.

Box 37-1 Lymphatic Malformation Stages of de Serres et al

- I. Unilateral infrahyoid lesion
- II. Unilateral suprahyoid lesion
- III. Unilateral suprahyoid and infrahyoid lesion
- IV. Bilateral suprahyoid lesion
- V. Bilateral suprahyoid and infrahyoid lesion



Fig. 37-2 A, Preoperative appearance of a 5-year-old girl with a lymphatic malformation of the right side of her face. She is shown after several attempts at sclerotherapy with limited improvement. B, Wide exposure with preauricular incision, intraoperative nerve monitoring, and a visible lymphatic malformation. C, The specimen after partial excision. D, Postoperatively, the patient has improved facial contour and facial nerve preservation.

Patients with lymphatic malformations typically present at birth or in early infancy with a soft, spongy, painless mass that transilluminates. Some patients may be diagnosed prenatally with routine ultrasonography, whereas other diagnoses are delayed until later childhood when an inciting infectious or inflammatory event makes the lymphatic malformation obvious. Because the size of these lesions varies significantly, they are associated with a variety of presentations, ranging from completely asymptomatic to causing bony or soft tissue overgrowth and airway compromise. CT/MRI is used to characterize lesions and delineate relevant anatomic location.

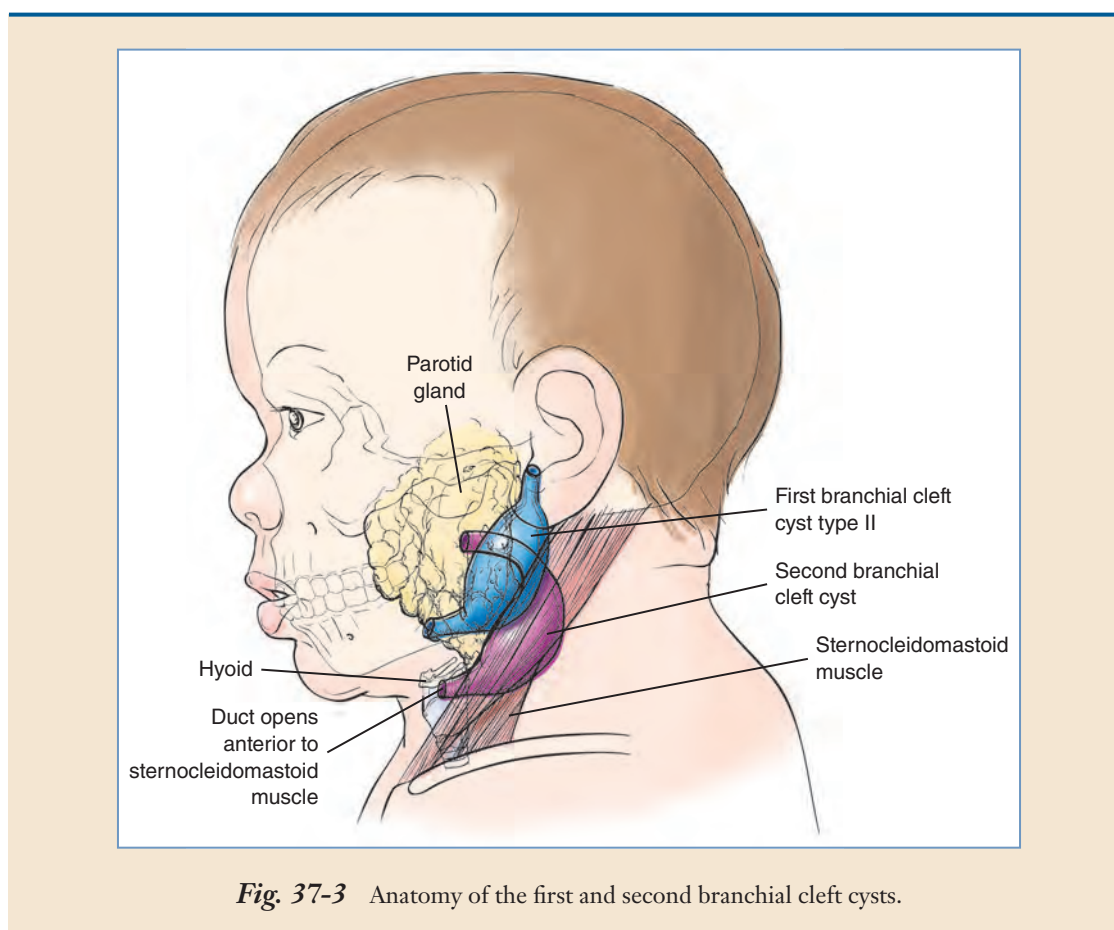
In the past, surgical excision was the treatment of choice for lymphatic malformations. Currently, sclerotherapy is more frequently used as first-line therapy for these lesions, particularly for macrocystic and mixed disease. Sclerotherapy is not as effective for microcystic disease. When surgical resection is attempted, intraoperative nerve monitoring may be useful during dissection, and subtotal resection may be necessary to preserve important structures (Fig. 37-2).

Branchial Cleft Lesions

The branchial arches and associated clefts and pouches develop in the fourth week of gestation. Initially there are six paired arches, but the fifth arch quickly involutes. The remaining arches are associated with an external cleft and an internal pouch. Each arch is lined internally by endoderm and externally by ectoderm and contains mesodermal elements, which give rise to nerve, arterial, and skeletal structures⁴ (Table 37-1).

Table 37-1 Branchial Arch and Associated Derivatives

Derivatives of Pharyngeal Folds	Arch Number	Cranial Nerve	Muscles	Skeletal	Artery
External auditory meatus	I. Mandibular	V. Trigeminal	Muscles of mastication, mylohyoid, anterior belly of digastric, tensor palatini, tensor tympani	Premaxilla, maxilla, Meckel's cartilage, squamous temporal bone, malleus, incus, sphenomandibular ligament	Maxillary
Neck	II. Hyoid	VII. Facial	Muscles of facial expression, buccinator, platysma, posterior belly digastric, stylohyoid, stapedius	Stapes, stylohyoid process/ligament, part of hyoid cartilage	Hyoid, stapedial
	III.	IX. Glossopharyngeal	Stylopharyngeus muscle	Part of hyoid cartilage	Common/internal carotid
	IV.	X. Vagus (Superior laryngeal)	Cricothyroid muscle, soft palate musculature (except tensor veli palatini)	Thyroid cartilage, parathyroids, epiglottis	Right subclavian, aorta
	VI.	X. Vagus (Recurrent laryngeal)	Laryngeal musculature	Cricoid, styloid, corniculate cartilage	Pulmonary, ductus arteriosus



Branchial cleft anomalies are one of the most common types of congenital neck lesion and include cysts, internal sinuses, external sinuses, and fistulas. The most common of these are *branchial cleft cysts*, which are congenital, epithelium-lined cysts that form on pathologic or incomplete fusion of two adjacent arches. These lesions are relatively common, composing as much as one third of all pediatric neck lesions. They arise in predictable locations that are dependent on which arch is involved (Fig. 37-3). No ethnic or gender association has been reported for these lesions.

Although these are congenital lesions, they are not always diagnosed at birth. Many patients with branchial cleft cysts are diagnosed in childhood or even adulthood when the cyst becomes inflamed, frequently after an upper respiratory tract infection. Patients may present with pain, swelling, erythema, dysphagia, dyspnea, drainage, otorrhea, or halitosis, depending on the cyst location. In addition to a history and physical examination, imaging is useful in diagnosing branchial cleft cysts. Ultrasonography is a first-line imaging modality to diagnose these cystic lesions, whether a cyst or sinus tract is appreciable on physical examination. CT/MRI may show further definition of the lesion and demonstrate its relationship to the surrounding structures.

First Branchial Cleft Cysts

First branchial cleft cysts are rare, composing less than 10% of all branchial arch lesions.⁵ There are several classifications of these lesions, including the *Work classification*, which describes these lesions as type I and type II.⁶ Type I lesions are of ectodermal origin and thus are lined with squamous epithelium. Their location parallels the external auditory canal, with a possible fistula opening in the postauricular or pretragal region. The facial nerve is not typically involved (Fig. 37-4). Type II lesions are more common than type I lesions. Both are of ectodermal and mesodermal origin and therefore may contain squamous epithelium and cartilage. Type II lesions are typically found between the external auditory canal and neck, superior to the hyoid bone. The facial nerve and parotid gland are more intimately associated with these lesions than with type I lesions (Fig. 37-5). If possible, the treatment of both of these lesions is surgical excision if inflammation is not present. Wide exposure through a preauricular parotidectomy incision is preferred for adequate visualization and avoidance of the facial nerve (Fig. 37-6). Superficial parotidectomy may be necessary to completely remove these lesions and preserve the facial nerve. Intraoperative nerve monitoring is often used, because the normal anatomy may be distorted as a result of the lesion.



Fig. 37-4 A type I lesion of the first branchial sinus tract presenting as a preauricular pit. At excision, the sinus tract extends to the helical crus, where a segment of cartilage must be resected to prevent recurrence.

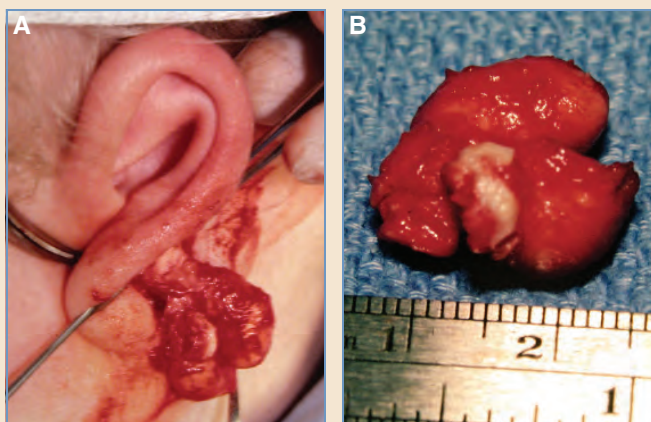


Fig. 37-5 A, A cervicoauricular cyst and sinus tract extending to the external auditory canal. B, A segment of the cartilage must be resected with the sinus tract to prevent recurrence.

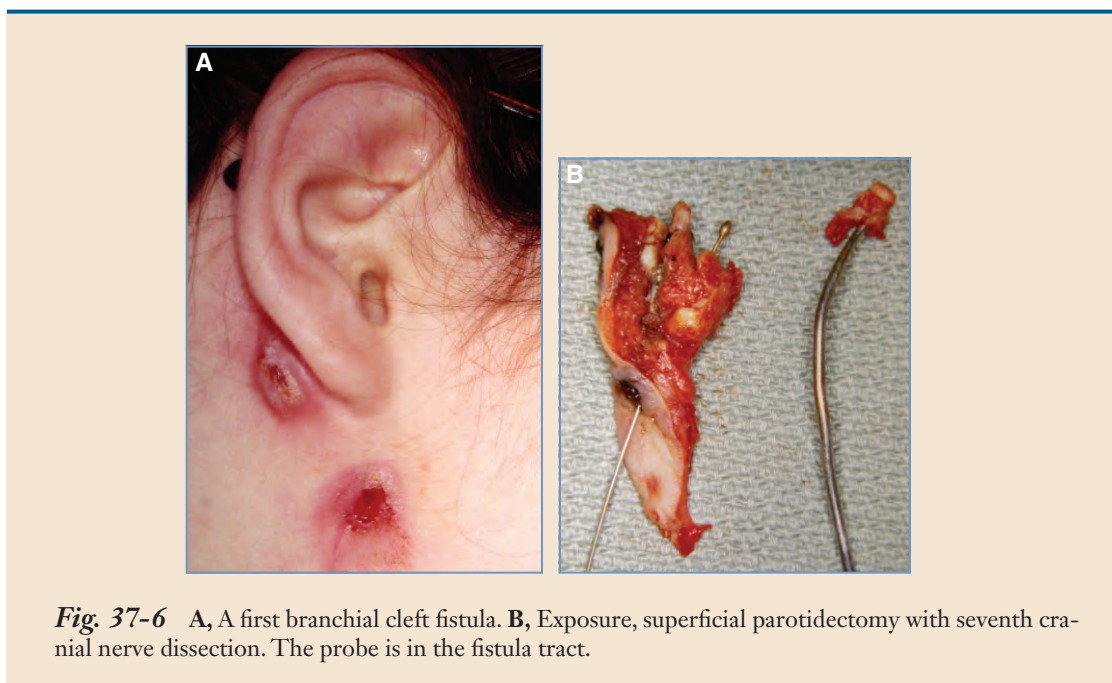


Fig. 37-6 A, A first branchial cleft fistula. B, Exposure, superficial parotidectomy with seventh cranial nerve dissection. The probe is in the fistula tract.

Second Branchial Cleft Cysts

Second branchial cleft cysts are by far the most common branchial pathology, accounting for 90% of these lesions. The second branchial arch contributes the future hyoid bone, and these lesions are located in the neck, inferior to the hyoid. If a cutaneous fistula is present, the opening is typically found along the anterior border of the sternocleidomastoid muscle (SCM). It then travels deep to the platysma and deep and posterior to the submandibular gland, between the internal and external carotid arteries, and over the hypoglossal and glossopharyngeal nerves to terminate in the tonsillar fossa (Fig. 37-7). A cyst may be present anywhere along this path. Historically, these cysts have been described by anatomic location, according to the Bailey classification^{7,8} (Box 37-2). The isolated second branchial arch cyst typically presents laterally in the neck below the angle of the mandible without a sinus tract and in most patients does not present until adulthood (Fig. 37-8).

Although anomalies of the remaining branchial arches exist, they are exceedingly rare, accounting for less than 2% of branchial cleft cysts. Because of their location, these lesions may be confused with suppurative thyroiditis or other thyroid pathology.⁹

Third Branchial Cleft Cysts

Third branchial cleft cysts begin at the piriform aperture and then travel superior to the superior laryngeal nerve and between the hypoglossal and glossopharyngeal nerves. Most often, they are found posterior to the SCM in the posterior cervical space. They are more commonly found on the left side.

Fourth Branchial Cleft Cysts

Fourth branchial cleft cysts begin at the piriform sinus but travel inferior to the superior laryngeal nerve and down the tracheoesophageal groove.¹⁰ Third and fourth branchial cleft cysts typically cannot be distinguished radiographically. Differentiation is largely based on the relationship to the superior laryngeal nerve and thus is made intraoperatively.

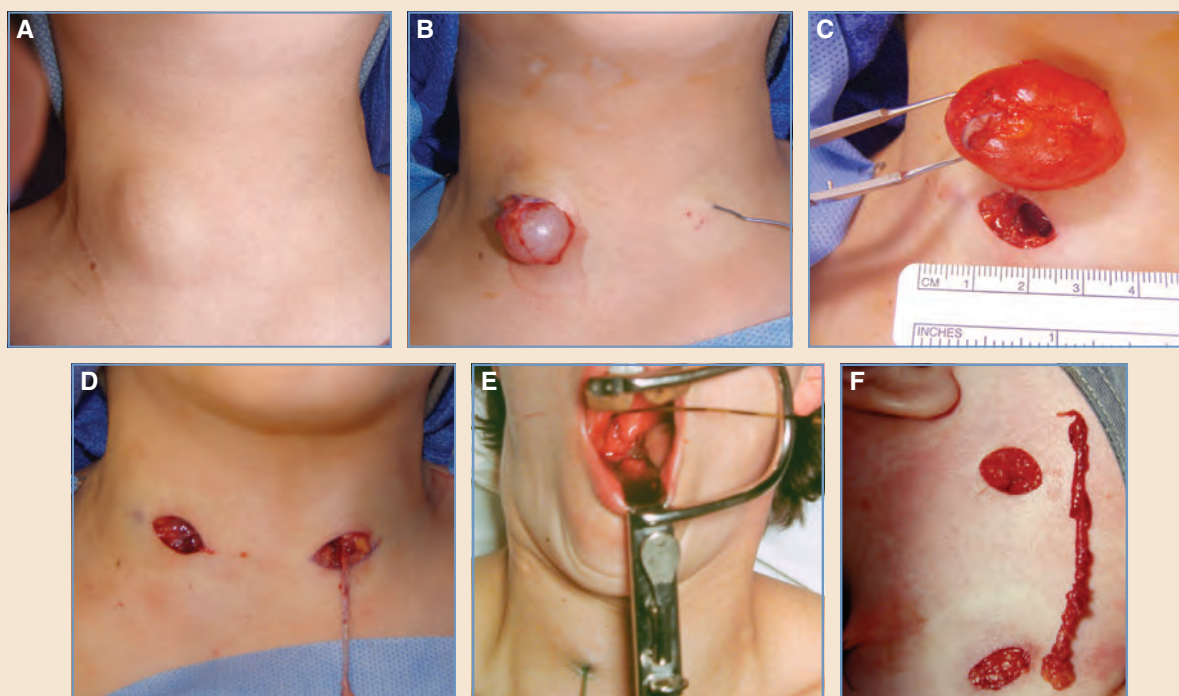


Fig. 37-7 A-D, This bilateral second branchial cleft cyst and sinus presented with the cyst alone in the right side of the neck but with the sinus tract opening in the typical location at the anterior border of the SCM at the junction of the mid and lower third of the muscle. Here, it was resected through a single incision. E and F, The entire embryologic tract can be seen with canalization of the tract extending into the pharynx. This was excised through the often-described *stairstep incisions*.



Fig. 37-8 An isolated second branchial cleft cyst in a young child.

Box 37-2 Bailey Classification of Second Branchial Cleft Cysts

Type I. Most superficial; lies deep to the platysma and anterior to the SCM

Type II. Most common; lies anterior to the SCM, lateral to the carotid space, posterior to the submandibular gland, and adherent to the great vessels

Type III. Extends medially between the external carotid artery and internal carotid artery to the lateral pharyngeal wall

Type IV. Lies within the pharyngeal mucosal space

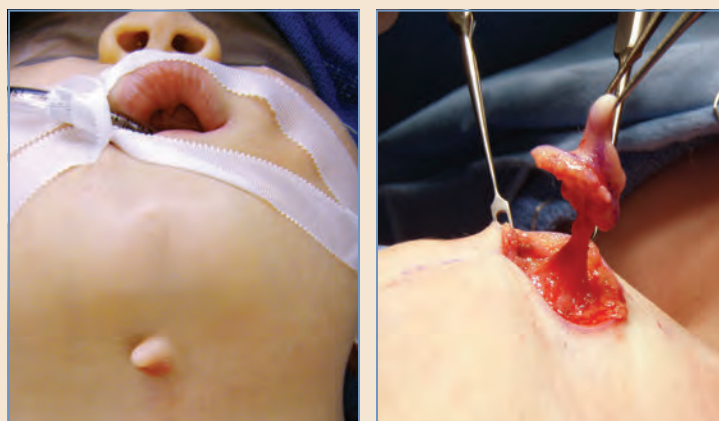


Fig. 37-9 This midline branchial vestige was not associated with a sinus tract but had an underlying amorphous cartilage vestige and a fibrous tract to muscle in the midline.

Treatment

Surgery is the mainstay of treatment of the second, third, and fourth branchial cleft cysts. Horizontal stairstep incisions placed in natural neck creases may be used to identify and resect the cyst and/or fistula tract in its entirety, although resection through a single incision is possible in some patients (see Fig. 37-7). The surgeon must be careful to avoid cranial nerves IX through XII during this dissection, particularly when small stairstep incisions are used and visibility is limited. Patients with an inflamed lesion can be given a course of antibiotics, with or without lesion aspiration, to decrease the local inflammation in preparation for surgical excision.

Branchial Cartilaginous Remnant or Cartilaginous Rests

Branchial cartilaginous remnants or *cartilaginous rests*, although considered lesions of the branchial apparatus, are far less common than branchial cleft cysts and sinuses. They are cervical deposits of cartilage most frequently found at the anterior border of the SCM and can be unilateral or bilateral. They are usually diagnosed in infancy. Affected patients present with asymptomatic masses in the neck that contain palpable cartilage within their substance. Treatment is surgical excision¹¹⁻¹³ (Fig. 37-9).

Thyroglossal Duct Cysts

Thyroglossal duct cysts (TDC) are the second most common congenital neck lesions after branchial cleft cysts. The thyroid gland begins developing in the third week of gestation; it starts as an epithelial invagination at the foramen cecum and makes its descent anterior to the hyoid to its adult position anterior to the larynx. Thyroid gland development is completed by the eighth week of gestation, and the tract is normally obliterated in the following 2 weeks. However, if epithelium persists at any point along the tract, a TDC may form.¹⁴

Approximately half of patients with TDCs present before 20 years of age.⁵ A typical presentation of these lesions is a child with an asymptomatic midline neck mass overlying the hyoid, but these lesions have the potential to become inflamed or infected (Fig. 37-10). A history and physical examination are generally adequate to diagnose a TDC. They can best be seen with the patient's neck extended and should elevate with swallowing or tongue protrusion. The patient's normal

thyroid gland should be examined to verify its presence. If further imaging is desired for evaluation of the lesion itself or the native thyroid gland, ultrasonography is the modality of choice. When a normal thyroid gland is detected on physical examination or imaging, a thyroid scan is not required. If, however, a normal thyroid gland is not detected, a TDC must be differentiated from ectopic thyroid tissue. As much as 10% of ectopic thyroid tissue is found in the neck, and in most of these patients, it is the only functioning thyroid gland.

Management of these lesions is surgical excision with the Sistrunk procedure. Sistrunk¹⁵ first described this procedure in 1920. It involves a small, transverse incision over the cyst, followed by excision of the cyst itself along with the tract from the tongue base to the thyroid gland and the central third of the hyoid bone¹⁶ (see Fig. 37-10). The Sistrunk procedure has repeatedly shown a lower incidence of recurrence than less aggressive techniques such as cyst-only excision.¹⁵

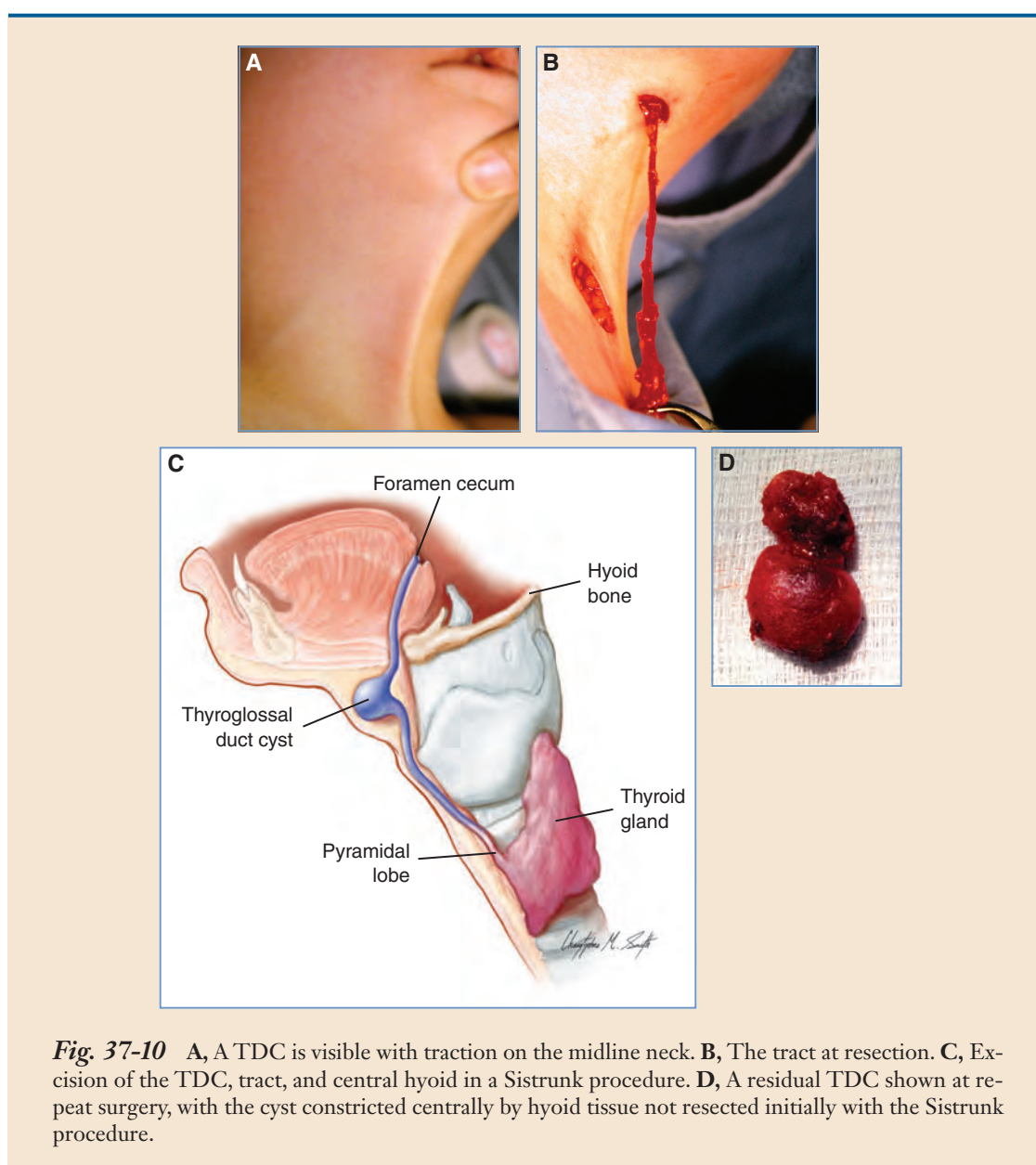


Fig. 37-10 A, A TDC is visible with traction on the midline neck. B, The tract at resection. C, Excision of the TDC, tract, and central hyoid in a Sistrunk procedure. D, A residual TDC shown at repeat surgery, with the cyst constricted centrally by hyoid tissue not resected initially with the Sistrunk procedure.

Dermoid Cysts

The term *dermoid cysts* may be used to describe several different lesions, including dermoid cysts, epidermoid cysts, and teratomas. These lesions differ in their composition and embryologic origin. Epidermoid cysts arise from ectodermal tissue, dermoid cysts from ectodermal and mesodermal tissue, and teratomas from all three germ layers. Dermoid cysts are commonly encountered congenital masses, accounting for 60% of all facial cysts.¹⁷ These cysts are frequently located along sites of embryologic soft tissue or bony fusion. In the head and neck region, this includes periorbital, nasal, intraoral, anterior and posterior fontanelles, and postauricular sites of formation (Fig. 37-11). They are lined with stratified squamous epithelium and may contain hair follicles and other dermal appendages.

Patients with dermoid cysts typically present with a painless, firm, palpable mass that does not transilluminate or change with crying. However, they may become infected, in which case patients may present with signs of infection and inflammation. For dermoid cysts not occurring in the midline, diagnosis may be based on clinical examination, and no further imaging is required. Midline nasal dermoid cysts require investigation into possible intracranial communication. (See Chapter 35 for further discussion of nasal dermoid cysts.)

Dermoid cysts are treated by complete surgical excision, preventing cyst rupture when possible. Underlying adherent periosteum should be included in the resection specimen. Incomplete excision is acceptable in patients in whom the cyst is adherent to vital structures.¹⁸ Patients should be monitored for cyst recurrence, especially in incompletely resected lesions.

Teratomas

Teratomas are rare germ cell tumors that are derived from all three germ layers: ectoderm, mesoderm, and endoderm. They develop from aberrant pluripotent cells that stray in the fourth to fifth week of gestation. They may be classified as mature or immature, depending on the tissue type present. Mature teratomas account for most pediatric teratomas. Mature teratomas contain well-differentiated cells and structures, and they are associated with elevated levels of alpha-fetoprotein. Immature teratomas are poorly differentiated and contain embryonal elements. They have a significantly higher risk of malignant transformation and are also associated with elevated beta-human chorionic gonadotropin.

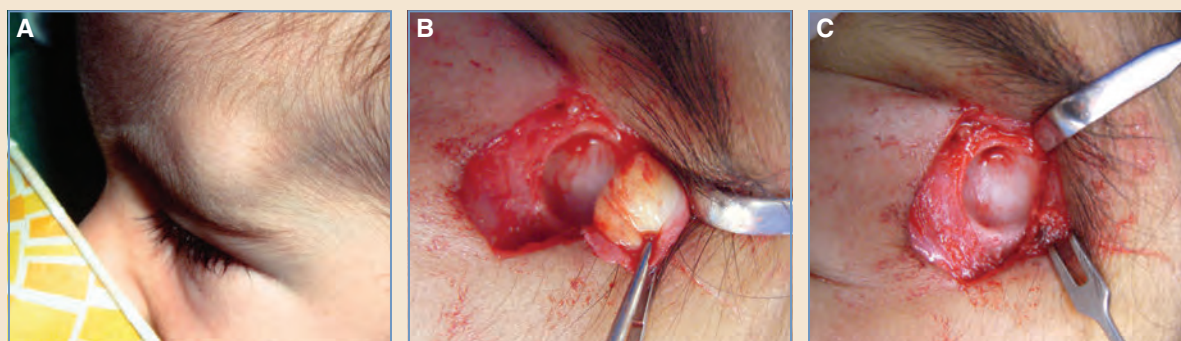


Fig. 37-11 An external angular dermoid cyst located at the zygomaticofrontal suture. **A**, The most common presentation is either a fixed or mobile cyst at the lateral orbit. **B** and **C**, At excision, this cyst is positioned in a concavity in the orbital bone at the frontozygomatic suture.

Head and neck teratomas account for 2% to 5% of all germ cell neoplasms and include cervical and craniofacial teratomas.¹⁹ They are frequently diagnosed with prenatal ultrasonography. They appear as large semicystic lesions and may be difficult to distinguish from a lymphatic malformation or branchial cleft cyst. Polyhydramnios may also be seen on prenatal ultrasonography as a result of the fetus's impaired ability to swallow because of the mass effect of the lesion. Diagnosis can be further investigated with fetal MRI.

Treatment

Treatment of these lesions, whether mature or immature, is surgical excision. The EXIT procedure may be performed for lesions diagnosed prenatally to secure the patient's airway in preparation for tumor excision. Tumor excision is ideally performed in the first 48 hours of life or after diagnosis to prevent complications associated with airway impingement by the lesion itself or loss of airway with a hemorrhagic event. For aggressive, immature lesions, chemotherapy may be considered. The prognosis is excellent for patients who undergo successful airway stabilization and tumor resection (Fig. 37-12).

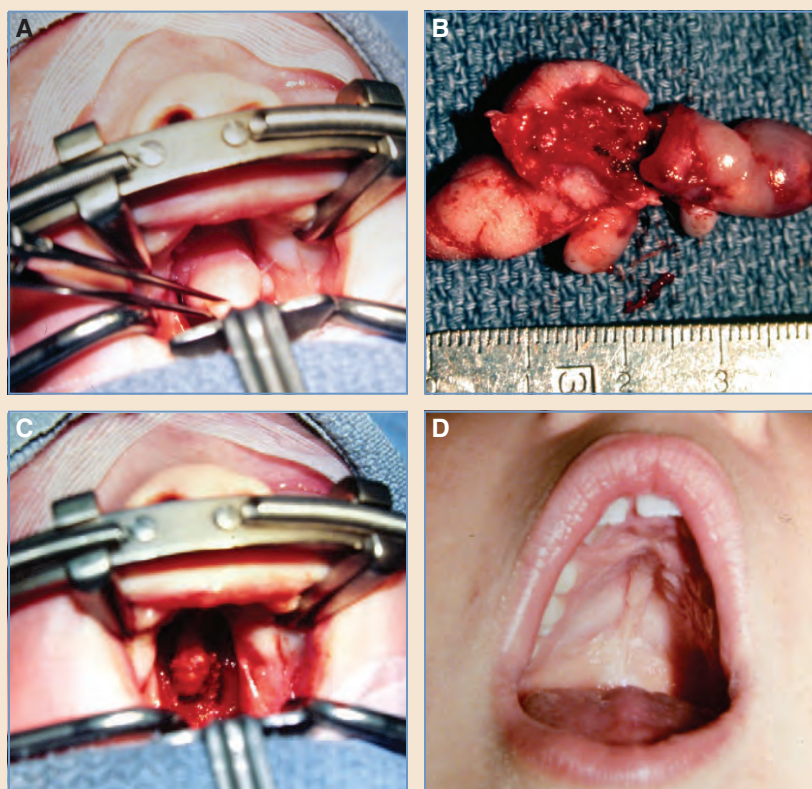


Fig. 37-12 A, A teratoma of the vomer, resulting in a cleft palate. B, The resected teratoma. C, The cleft of the secondary palate. D, The repaired palate 5 years after repair.

Heterotopic Neuroectodermal Tissue

Heterotopic neuroectodermal tissue comprises a group of lesions that includes encephaloceles and gliomas. Both present with craniofacial masses in various locations—most commonly occipital and anterior (sincipital) (in the glabellar region). Although benign, these lesions may be disfiguring and are treated by surgical excision to prevent further craniofacial distortion. (See Chapter 39 for further discussion of encephaloceles and gliomas.)

Congenital Muscular Torticollis and Pseudotumor of Infancy

Congenital muscular torticollis (CMT) and *pseudotumor of infancy* (POI) are musculoskeletal conditions characterized by unilateral shortening of the SCM with or without palpable intramuscular mass. As a result, contralateral face/chin rotation and ipsilateral head tilt occur.^{20,21} It is unclear whether these two conditions are pathologically distinct, but most commonly POI refers to an intramuscular neck mass that develops by 6 weeks of age, whereas CMT results after 6 weeks. The cause of CMT and POI is not well understood. Theories have been published proposing intrauterine fetal positioning, birth trauma, and familial inheritance as the causes of the disorder.²² The diagnosis of these conditions may be made clinically. If further confirmation is needed, ultrasonography is the imaging modality of choice and may demonstrate a fibrotic lesion of heterogeneous echogenicity. Fine-needle aspiration with interstitial fibrosis may also be performed to confirm the diagnosis, but this is unnecessary in most cases.²⁰ Initial management consists of massage and passive range-of-motion exercises performed at home by the child's family or if this is unsuccessful by physical therapists. If successful, some improvement should be noted by 8 weeks, but on average the duration of therapy for maximal benefit is 4 months. In most cases (90%), physical therapy is successful, but if no improvement is noted, surgical intervention consisting of open or endoscopic disinsertion of the sternal and clavicular heads of the SCM should be considered. Varying success with botulinum toxin injection directly into the affected muscle has been described.^{1,23-25}

Cervical Thymic Cysts

Cervical thymic cysts are rare congenital pediatric neck masses. Beginning in the third week of gestation, the thymus gland develops from the third branchial pouch and descends into its final resting place in the mediastinum. Cervical thymic cysts can occur with implantation of thymic tissue anywhere along this tract, from the angle of the mandible to the mediastinum. Regardless of location, they frequently have some connection to the mediastinum.²⁶ These cysts are usually diagnosed in patients younger than 20 years of age.²⁷

Patients typically present with a painless lateral neck mass, usually on the left side. In unusual cases these lesions may be sufficiently large so that they cause stridor, dyspnea, hoarseness, and dysphagia. On physical examination, they appear similar to branchial cleft cysts both in appearance and location. Ultrasonography may reveal a unilateral, unilocular, or multilocular cyst in the lateral neck and inferior extension into the mediastinum. Surgical excision is the treatment of choice for these lesions. Diagnosis relies on a tissue sample obtained at surgical excision of the cyst.

CONGENITAL OR ACQUIRED LESIONS

Ranulas

Ranulas are submucosal extravasation of sublingual salivary glands. They may be congenital or acquired. Congenital ranulas may result from an imperforate sublingual salivary gland duct and frequently resolve spontaneously. Acquired ranulas are often the result of trauma to the duct and are more likely to recur if no intervention is performed. They may be classified based on anatomic location as either simple or plunging ranulas. Simple ranulas are confined to the floor of mouth, whereas plunging ranulas extend through a natural separation in the mylohyoid muscle into the neck.²⁸ Although typically asymptomatic, these lesions may cause tongue elevation or dysphagia if they are of sufficient size. On physical examination, simple ranulas appear as a fluctuant, blue-tinged dome in the floor of mouth that will transilluminate. Plunging ranulas may appear as a soft, fluctuant neck mass of varying size. Various imaging modalities have been used to diagnose these lesions, including ultrasonography, CT, and MRI. The lesion contents may be aspirated for diagnostic purposes and will yield amylase-rich fluid. Treatment for ranulas includes observation, marsupialization, incision and drainage, sclerotherapy, sublingual salivary gland lobe resection, and radical resection of the sublingual gland. Of these, resection of the entire sublingual gland through either the intraoral or transcervical approach is most likely to result in the lowest chance of recurrence.^{29,30}

ACQUIRED LESIONS

Infectious Lesions

Infectious causes of acquired head and neck masses can create acute enlargement of the regional lymph nodes or salivary glands. Most acquired head and neck masses in the pediatric population are inflammatory or infectious. Infections may be bacterial, viral, or fungal. In investigating a pediatric head or neck mass, surgeons should obtain a thorough history from the patient or caregivers; special attention should be paid to the temporal relationship of mass and infection, recent travel, exposure to animals, sick contacts, vaccination history, trauma, associated symptoms, and drug use. Although a clinical diagnosis, laboratory tests, including cultures and imaging, may often help to determine a definitive diagnosis.

Sialadenitis

Sialadenitis is the inflammation of any of the salivary glands. Bacterial sialadenitis is most common in infants younger than 2 months of age. Infection is typically polymicrobial, with *Staphylococcus aureus* most often isolated. *Haemophilus influenzae*, *Peptostreptococcus*, *Streptococcus pneumoniae*, *Escherichia coli*, *Bartonella henselae*, and *Bacteroides* are also culprits. Viral sialadenitis may be caused by mumps, Epstein-Barr virus, cytomegalovirus, adenovirus, influenza, herpesvirus 6, parainfluenza, coxsackievirus, or human immunodeficiency virus. Granulomatous diseases may affect the salivary glands through the periglandular lymph nodes. Atypical mycobacteria, tuberculosis, actinomycosis, and sarcoidosis are causative organisms.

If infectious sialadenitis is suspected, in addition to the information previously listed, the temporal relationship of symptoms to eating should be determined. On examination the gland should be palpated for stones, and an attempt should be made to massage the gland, obtain fluid

for culture, or identify ductal obstruction. In the pediatric patients, ultrasonography is the first-line imaging modality to assess salivary gland masses. If an abscess is identified, fine-needle aspiration may be performed to decompress the gland and to obtain fluid for culture. Radiographs may facilitate visualization of sialoliths, although many are radiolucent. CT scans will help to identify ductal stones, strictures, abscesses, and tumors. Sialography involves injecting contrast retrograde into the duct to detect pathology. It can provide useful information but is difficult to perform in children. Sialendoscopy is also difficult to perform in children but allows visualization of ductal anatomy and pathology. Treatment of sialadenitis includes antibiotics when indicated, aspiration or incision and drainage of the abscess, sialogogues, warm compresses, and hydration.

Lymphadenitis

Lymphadenitis is inflammation of the lymph nodes. Bacterial lymphadenitis is more commonly seen in children younger than 4 years of age. Submandibular lymph nodes are the most frequently affected and are seen in more than half of patients. Organisms causing bacterial adenitis include *S. aureus*, *S. pyogenes*, *S. agalactiae*, *B. henselae*, and *E. coli*. Cat scratch disease is caused by *B. henselae*, and inoculation usually occurs through a cat bite or scratch. Most cases are found in patients younger than 20 years of age. Viral lymphadenitis is most often seen when reactive lymphadenopathy occurs after a viral upper respiratory tract infection. Causative organisms include adenovirus, rhinovirus, coxsackievirus, and Epstein-Barr virus. Granulomatous diseases may be caused by organisms such as atypical mycobacteria, tuberculosis, actinomycosis, and sarcoidosis. Epstein-Barr virus is associated with infectious mononucleosis, which frequently affects adolescents. Patients classically present with the triad of lymphadenopathy, acute pharyngitis, and fever. Splenomegaly may occur, and patients should refrain from participation in contact sports and other activities to prevent splenic rupture.

Inflammatory Lesions

Noninfectious inflammatory pathology can cause lymphadenopathy in the head and neck region. Causes of noninfectious inflammatory disease include vasculitis, autoimmune disease, and drug side effects.

Kawasaki Disease

Kawasaki disease is a vasculitis of medium-sized vessels that typically affects patients younger than 5 years of age. Diagnosis of Kawasaki disease is made clinically with 5 days of fever plus 4 days of the following symptoms: cervical lymphadenopathy, conjunctival injection, an erythematous/edematous rash on the hands and feet, strawberry tongue/lip fissures, and a polymorphous rash. Without treatment, about 25% of children will also develop coronary artery aneurysms. Treatment consists of aspirin and gamma-globulin therapy, with the goal of decreasing inflammation, symptoms, and complications.

Sjögren Syndrome

Sjögren syndrome is an autoimmune disease targeting the salivary and lacrimal glands. The major symptoms are keratoconjunctivitis, sicca, and xerostomia. In children, the most common symptom at presentation is salivary gland enlargement. Diagnosis may be aided with laboratory tests, including an elevated erythrocyte sedimentation rate, positive antinuclear antibody, positive rheumatoid factor, positive SS-A and SS-B, and hypergammaglobulinemia. Treatment is mainly aimed at symptom alleviation.

Sarcoidosis

Sarcoidosis is a systemic inflammatory disease of unknown cause. Although sarcoidosis typically affects individuals in their twenties, it may be seen in any age group. Symptoms vary depending on which organs are affected by the disease. Cervical lymphadenopathy is the most common head and neck manifestation of sarcoidosis. Most patients require only symptomatic treatment. For more severe disease, corticosteroids or other medical therapies are used.

Juvenile Recurrent Parotitis

Juvenile recurrent parotitis or *recurrent parotitis of childhood* is an inflammatory disorder of the parotid gland. It is most commonly seen in children 3 to 6 years of age. Although its cause is unknown, the disorder is characterized by pain and swelling of one parotid gland and associated fever and malaise, with one to five recurrences per year. Although each episode tends to be unilateral, both parotid glands are usually involved. Episodes typically last less than 1 week and are self-limited. On physical examination, patients present with parotid swelling and pain. Purulent secretions may be expressed from the gland. Diagnosis is supported by sialiectasis on sialography.²⁸ Treatment is generally supportive, but ductal irrigation may reduce the number of recurrences.¹⁴

Drug-Induced Lymphadenopathy

Drug-induced lymphadenopathy can be caused by a variety of medications, including anticonvulsants, antithyroidals, isoniazid, hydralazine, allopurinol, and phenytoin. In most patients, adenopathy resolves after the causative drug is discontinued.

NEOPLASTIC LESIONS

Cervical Lymphadenopathy

Cervical lymphadenopathy most likely has an inflammatory or infectious cause. However, 1.5 in 10,000 children younger than 20 years of age will be diagnosed with cancer, and 5% of neoplasms in children manifest in the head and neck region.^{9,31,32} Although most neoplasms in children are benign, as many as 10% are malignant.¹ Cervical lymphadenopathy that does not respond to initial intervention must have a workup to rule out malignancy. A rule of thumb is that a lymph node larger than 2 cm for longer than 2 months should undergo a biopsy procedure unless an infectious cause has been documented.

Lymphoma

Lymphoma is the most common malignancy in children, followed by retinoblastoma, rhabdomyosarcoma, neuroblastoma, thyroid carcinoma, and melanoma.¹⁴ In diagnosing these lesions, invasive procedures and imaging may require sedation. An effort should be made to minimize ionizing radiation and the need for sedation. Multiple procedures or imaging studies should be combined whenever possible.

Improvements in the survival rates of children with cancer have been significant, with mortality decreasing by more than 50% from 1975 to 2010.^{33,34} Children with a cancer diagnosis should generally be evaluated and treated in pediatric cancer centers, because this has shown a survival benefit in multiple different malignancies.³³ In this setting pediatric plastic surgeons may be asked to aid with tumor diagnosis by performing open biopsy procedures and possibly tumor extirpation. Usually, however, plastic surgeons perform complex reconstruction after resection of the lesion. Reconstruction can be challenging, with large hard and soft tissue defects to recapitulate, often in irradiated tissues (Fig. 37-13).

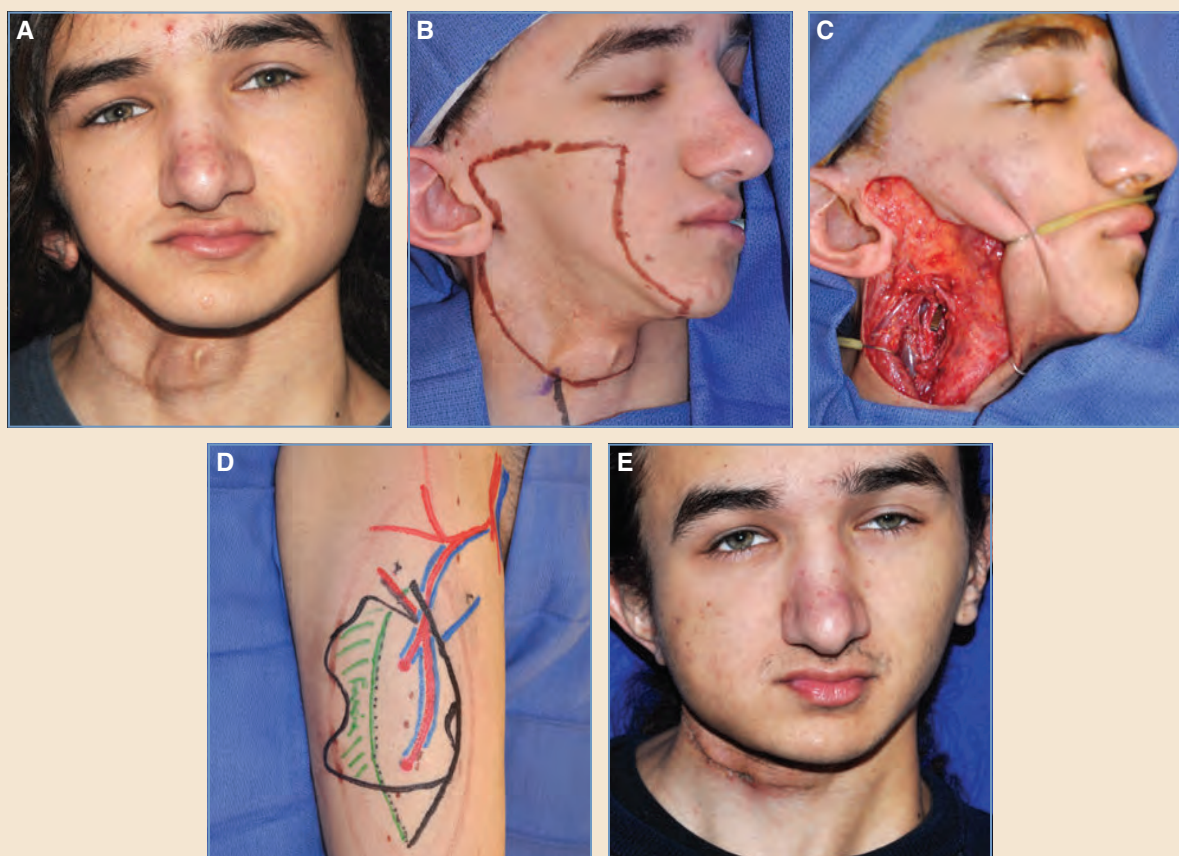


Fig. 37-13 A, The preoperative appearance of an 18-year-old boy who underwent rhabdomyosarcoma excision and radiotherapy to the right side of the neck when he was a child. B, The outline highlights the area to be reconstructed. C, An extended preauricular incision made for adequate exposure of recipient vessels in the neck and to optimize the aesthetic appearance. D, A free anterolateral thigh flap used to restore facial volume. E, Postoperative appearance with improved facial symmetry at 4 months.

Lymphoproliferative Conditions and Histiocytoses

Lymphoma is not only the most common malignancy in children, it is also the most common pediatric head and neck cancer.^{14,35} Lymphomas represent a diverse group of neoplasms of the lymphoreticular system. They are classified primarily as either *Hodgkin lymphoma* (HL) or *non-Hodgkin lymphoma* (NHL).

Although not well understood, there appears to be an association between infection with the Epstein-Barr virus and multiple pediatric malignancies, including HL and NHL. Initially, patients with a history of Epstein-Barr virus were thought to have an increased epidemiologic risk of developing lymphoma.^{36,37} Subsequent studies have demonstrated high rates of Epstein-Barr virus positivity among tumor cells.³⁷ This is particularly true in the endemic form of Burkitt lymphoma.

Hodgkin Lymphoma

HL accounts for 6% of pediatric malignancies. It has a bimodal age distribution, which peaks in patients 15 to 40 years of age and in those older than 60 years of age. Clinically, it may be classified by age group: childhood (younger than 14 years of age), young adult (15 to 34 years), and older adult (55 to 74 years).³⁸ In industrialized nations, the incidence of HL is highest in adolescents 15 to 19 years of age, whereas in the developing world, the incidence is higher in childhood.³⁸ *Reed-Sternberg cells* are identified histologically as large, multilobular/multinucleated cells and are pathognomonic for HL.

Clinically, HL typically presents as an asymptomatic cervical or supraclavicular “rubbery” mass. Patients may complain of so-called “B symptoms” or constitutional symptoms, which include a fever higher than 38° C for 3 consecutive days, weight loss of 10% in the 6 months preceding presentation, and drenching night sweats. Patients may have been seen and treated previously for a presumed inflammatory or infectious cause.

Preoperative treatment includes a thorough history and physical examination. Standard laboratory tests include a complete blood cell count and metabolic panel, serum copper and serum ferritin levels, erythrocyte sedimentation rate, and C-reactive protein. Imaging may be performed and can include a chest radiograph, neck/chest CT, abdomen/pelvis CT or MRI, and a PET scan. Further intervention includes obtaining a bone marrow biopsy and CSF sampling.

Unlike solid tumors, staging for HL is based on the anatomic location of the disease. The Ann Arbor staging system with Cotswold modification is currently used to stage patients diagnosed with HL and to monitor their progress³⁹ (Table 37-2). Previously, surgical staging was performed with an exploratory laparotomy for lymph node biopsy or splenectomy. Current imaging techniques can provide information about abdominal lymphatic and extralymphatic involvement.

Treatment for HL is multimodal with chemotherapy and radiotherapy. Because of the high survival rates of this disease, it is particularly important to prevent the iatrogenic effects of therapy. Treatment protocols aimed at minimizing chemotherapy and radiation doses have been developed for this purpose.

The prognosis for children and adolescents with HL is excellent, with a disease-specific and overall survival rate of more than 90%.^{40,40a} Factors associated with a poorer prognosis include advanced stage, the presence of B symptoms, bulky disease or extranodal involvement, leukocytosis, an elevated erythrocyte sedimentation rate, anemia, and male sex.⁴⁰

Table 37-2 Cotswold Modification of the Ann Arbor Staging System

Stage	Area of Involvement
I	Single lymph node group
II	Multiple lymph node groups on the same side of the diaphragm
III	Multiple lymph node groups on both sides of the diaphragm
IV	Multiple extranodal sites or lymph nodes and extranodal disease
X	Bulk >10 cm
E	Extranodal extension or a single, isolated site of extranodal disease
A/B	B symptoms: weight loss >10%, fever, drenching night sweats

Non-Hodgkin Lymphoma

NHL is a diverse group of diseases of lymphoid origin that can arise from T cells, B cells, or natural killer cells. It accounts for approximately 7% of malignancies in patients younger than 20 years of age, and the incidence is highest in adolescents, similar to HL.⁴¹ NHL is divided into several subtypes based on cell of origin, including Burkitt lymphoma, diffuse large B-cell lymphoma, lymphoblastic T- or B-cell lymphoma, and anaplastic large-cell lymphoma. Other subtypes such as follicular lymphoma and marginal zone lymphoma exist but are significantly less common. Histologically, a “starry sky” appearance is seen because of the numerous macrophages that have consumed the apoptotic cells. Of all of the various subtypes of NHL, Burkitt lymphoma is the most common.

In contrast to HL, patients with NHL are more likely to present with diffuse, extranodal disease. Extranodal disease is most frequently seen in the liver, lungs, and bone marrow. In addition to a history and physical examination, patients should undergo the laboratory workup and imaging studies described for HL. The most consistently used staging system for NHL is the St. Jude (Murphy) staging system⁴² (Table 37-3).

Treatment for NHL in children is primarily nonsurgical and consists of various chemotherapy regimens. Radiotherapy, although used to treat adult NHL, is not frequently used in children.

The prognosis for NHL has significantly improved during the past 25 years, with an event-free survival rate of more than 80%, regardless of subtype. The survival rate with Burkitt lymphoma is even higher. The presence of bone marrow or CNS involvement is a poor prognostic sign.³⁴

Burkitt Lymphoma

Burkitt lymphoma has been classified into three distinct clinical forms: endemic, sporadic, and immunodeficiency associated. All of these forms are associated with a translocation of the *c-myc* oncogene on chromosome 8. Endemic Burkitt lymphoma is found primarily in Africa, where it accounts for up to 50% of all childhood cancer. Its incidence is approximately 8 in 100,000, with the highest incidence in children 4 to 7 years of age. Males are affected twice as frequently as females. Tumor location is the jaw in more than half of cases, with abdominal tumors also seen but less common. Ninety-five percent of cases are associated with the Epstein-Barr virus in tumor cells.⁴³ Sporadic Burkitt lymphoma is found in North America and Western Europe. Its incidence is 3 in 1,000,000, accounting for about 3% of pediatric lymphoma in the region. The highest incidence is in children 6 to 12 years of age, with males more frequently affected than females. Tumor location is most frequently the abdomen.⁴⁴ In sporadic Burkitt lymphoma, the Epstein-Barr virus is associated with only 15% of cases. Immunodeficiency-associated Burkitt lymphoma is less common than the other two forms and less relevant in the pediatric population. It is seen in patients with HIV infection.

Sarcomas

Rhabdomyosarcoma

Rhabdomyosarcoma is the most common soft tissue sarcoma in children, representing 4.5% of all childhood malignancies.^{45,46} Of children affected, 35% present with a primary head and neck tumor. The orbit is the most common site involved in head and neck tumors, but multiple other sites throughout the region may also be affected. Rhabdomyosarcoma has a bimodal age distribution, with peaks at 2 to 6 years of age and 10 to 18 years of age. This is partly because there are two histologic subtypes: embryonal and alveolar. The embryonal subtype has a higher incidence in infants and is less common through childhood. Embryonal-subtype tumors are more frequently located in the head and neck region. In contrast, alveolar-subtype lesions have a higher incidence

Table 37-3 Staging System for Non-Hodgkin Lymphoma

Stage	Staging for Hodgkin Lymphoma	Staging for Non-Hodgkin Lymphoma
I	Only one lymph node region/structure involved Only a single extranodal site	Single tumor/nodal area involved, excluding the abdomen and mediastinum
II	Two or more lymph node regions/structures on the same side of the diaphragm, ± local, contiguous, extranodal spread	Single tumor with regional nodal involvement Two tumors/nodal areas ± regional nodal involvement on the same side of the diaphragm Primary gastrointestinal tumor grossly completely resected ± mesenteric nodal involvement
III	Lymph node regions or structures above and below the diaphragm are involved, ± spleen, ± extranodal site, or both	Two or more tumors/nodal areas on the opposite sides of the diaphragm All primary intrathoracic disease (mediastinal, thymic, pleural) Extensive primary intraabdominal disease Paraspinal or epidural tumors
IV	Disseminated involvement of extranodal sites	Bone marrow or central nervous system involvement

in older children and adolescents and more commonly affect the trunk and extremities. Most cases of rhabdomyosarcoma are sporadic but are associated with some familial syndromes, including Li-Fraumeni syndrome and neurofibromatosis.⁴⁵

Patients typically present with a firm, painless mass in the affected area. Although frequently asymptomatic, tumors, especially of the head and neck region, may exert a mass effect on surrounding structures and cause symptoms such as nasal obstruction, rhinorrhea, sinusitis, epistaxis, and cranial nerve palsy. A workup of these patients includes a history and physical examination, including a full cranial nerve examination. Laboratory studies should include a complete blood cell count, a complete metabolic panel, and a urinalysis. CT and MRI are the imaging modalities of choice for these tumors. A metastatic workup may be performed and includes a bone marrow aspirate, a CSF analysis, and a CT scan of the brain, lungs, and liver. A tumor biopsy and a sentinel lymph node biopsy should be performed to confirm the diagnosis and disease stage.

Rhabdomyosarcoma is staged before and after treatment. Pretreatment staging is a TNM staging system that considers the site and size of the primary tumor, the degree of tumor invasion, the nodal status, and the metastatic disease.^{45,47} After tumor excision, patients are assigned to clinical groups based on the completeness of tumor excision and metastasis^{45,47} (Box 37-3). The preoperative staging and postoperative clinical grouping allow patients to be risk stratified, which guides further treatment.

Treatment of patients with rhabdomyosarcoma is multimodal. After the diagnosis is confirmed with a biopsy sample, surgical resection should be attempted, with the goal of obtaining a circumferential margin of 0.5 cm around the tumor. Head and neck tumors do not require lymph node sampling of clinically or radiographically negative lymph nodes. In addition to surgical resection, patients receive adjuvant chemotherapy and some also receive radiotherapy.

The prognosis for patients with rhabdomyosarcoma is affected by a variety of factors. Overall, however, the 5-year survival rate is 67% for children younger than 15 years of age and 51% for those 15 to 19 years of age.³⁴

Box 37-3 Rhabdomyosarcoma Postresection Clinical Groups

- I. A: Localized tumor confined to the organ/muscle of origin, complete resection
B: Localized tumor infiltrating the outside site of origin but no lymph node involvement, complete resection
- II. A: Localized tumor with complete gross resection but residual microscopic disease
B: Regional disease in lymph nodes, complete resection
C: Regional disease in lymph nodes with complete gross resection but residual microscopic disease
- III. A: Localized tumor or regional disease with residual gross tumor after biopsy
B: Localized tumor or regional disease with residual gross tumor after resection
- IV. Distant metastases present

Box 37-4 Nonrhabdomyosarcoma Soft Tissue Sarcomas

- Fibrosarcoma
- Osteosarcoma
- Clear cell sarcoma
- Dermatofibrosarcoma protuberans
- Epithelioid sarcoma
- Malignant fibrous histiocytoma
- Hemangiopericytoma
- Malignant peripheral nerve sheath tumor
- Leiomyosarcoma
- Liposarcoma
- Chondrosarcoma
- Synovial sarcoma
- Desmoplastic small round cell tumor
- Alveolar soft part sarcoma
- Undifferentiated sarcoma

Nonrhabdomyosarcoma Soft Tissue Sarcomas

Nonrhabdomyosarcoma soft tissue sarcomas are a heterogeneous group of tumors that combined account for approximately 4% of pediatric malignancies⁴⁸ (Box 37-4). Although these tumors are rare, the most commonly encountered tumors in children are dermatofibrosarcoma protuberans, synovial sarcoma, malignant fibrous histiocytoma, malignant peripheral nerve sheath tumor, and fibrosarcoma.⁴⁹ In pediatric patients, those most commonly found in the head and neck region include malignant peripheral nerve sheath tumor, alveolar soft part sarcoma, and malignant fibrous histiocytoma. Most of these tumors occur sporadically, but some may be associated with an inherited syndrome. Children with Li-Fraumeni syndrome, neurofibromatosis type 1, and familial adenomatous polyposis are at significantly higher risk of developing a soft tissue sarcoma.

Fibrosarcomas

Fibrosarcomas are low-grade, locally aggressive tumors that arise from fibroblasts. They have a bimodal age distribution with two peaks: younger than 5 years of age and 10 to 15 years of age. These two peaks correspond to the two distinct forms of the disease.⁵⁰

Congenital/infantile fibrosarcoma occurs in the earlier of the two peaks and may present as a congenital lesion or in children younger than 2 years of age. Infantile fibrosarcoma is caused by a genetic translocation and is distinct from the childhood variant. Childhood fibrosarcoma occurs in the 5- to 10-year-old age group.⁵⁰ These lesions most commonly occur in the extremities but can also be found in the head and neck region. A workup includes a history and physical examination, with MRI for further tumor delineation. The treatment of choice is wide resection with generous surgical margins.⁵¹ Neoadjuvant chemotherapy may be used in some cases; adjuvant therapy is more controversial. The infantile form of the disease has a significantly better prognosis than the childhood form, with a 5-year survival rate of 84%, whereas the childhood form is more similar to adult fibrosarcoma, and the 5-year survival rate is 62%.⁵²

Dermatofibrosarcoma Protuberans

Dermatofibrosarcoma protuberans are low-grade, soft tissue sarcomas that most commonly occur on the trunk and extremities. An estimated 10% to 15% of these lesions occur in the head and neck region.⁵³ They may present as a congenital tumor or affect patients well into advanced age. About 6% to 20% of these diagnoses are in children.^{53,54} Although they have a low risk of metastasis, the lesions are tenacious with a high propensity to recur. On clinical examination, dermatofibrosarcoma protuberans have a highly variable appearance. Typically, they are asymptomatic, indurated nodular or plaque-like lesions with a firm consistency that are adherent to skin but not deeper structures.⁵⁵ Associated pigment changes may be present. The treatment of choice for these lesions is wide, local excision, with 2 to 4 cm margins to investing fascia or pericranium. Excision with smaller, more conservative margins has resulted in local recurrence of up to 60%.⁵⁵ Other risk factors for recurrence include histologic fibrosarcoma transformation, less than 1 mm from surgical margins, increased cellularity, a high mitotic rate, and age older than 50 years. Some advocate Mohs micrographic surgery for resection of this disease. Although this technique is controversial, advocates argue for its use, particularly in children in whom a 2 to 4 cm margin is quite large and for anatomic areas such as the head and neck. Adjuvant radiotherapy or imatinib may be used for persistently positive margins or residual microscopic disease (Fig. 37-14).

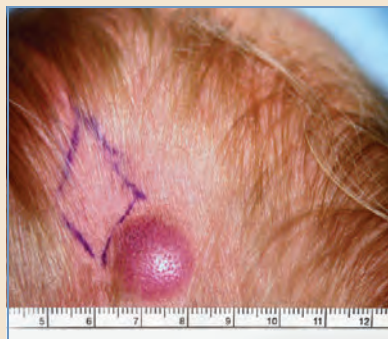


Fig. 37-14 Congenital dermatofibrosarcoma protuberans presenting as a firm reddish mass adjacent to but not overlying the anterior fontanelle. After confirmation, the adjacent scalp was expanded, and a wide resection and flap repair were carried out.

Synovial Sarcoma

Synovial sarcoma is a high-grade, spindle cell neoplasm that affects about 0.7 per million in the pediatric population.⁴⁰ It frequently occurs in close proximity to joints, joint capsules, tendon sheaths, and bursas. Most cases occur in the extremities, but it has also been reported in the head and neck. Patients with a synovial sarcoma usually present with slow-growing, painless tumors in the deeper soft tissues where they arise. Symptoms may arise from a mass effect on nearby structures. Imaging, including CT/MRI, is typically performed before a biopsy sample is obtained. Surgery with wide local excision is the mainstay of treatment for synovial sarcoma. Adjuvant radiotherapy may be used, but the use of chemotherapy is controversial. Overall survival rates of 70% at 5 years and 65% at 10 years have been reported in pediatric patients.⁵⁶ The prognosis is negatively affected by large tumor size (larger than 5 cm), tumors located in the lower extremity, and highly invasive tumors.⁵⁶

Carcinomas

Thyroid Carcinoma

Thyroid cancer in children is rare, accounting for 3% of pediatric cancer diagnoses.⁵⁷ However, it is the most common pediatric endocrine malignancy and accounts for approximately 5% of pediatric head and neck malignancies. Risk factors for developing thyroid cancer as a child include exposure to ionizing radiation, female sex, a personal or family history of thyroid disease, and iodine deficiency.⁵⁷⁻⁵⁹ There are several types of thyroid carcinoma, including papillary, follicular, and medullary. Papillary and follicular carcinoma originate in the follicular cells of the thyroid, and medullary carcinoma originates in the C cells. Although other subtypes exist, they are not typically found in children.

Papillary thyroid carcinoma is the most common type of thyroid cancer in both children and adults. In children it accounts for more than 90% of cases.¹⁴ Papillary thyroid carcinoma is frequently bilateral and multifocal; it usually metastasizes through the lymphatics, leading to regional disease. *Follicular thyroid carcinoma* is more often unifocal and metastasizes through hematogenous spread. Metastases are most commonly found in the lung and bones. *Medullary thyroid carcinoma* is often multifocal and bilateral. It is frequently associated with multiple endocrine neoplasia syndromes inherited in an autosomal dominant pattern.

Patients may or may not present with a thyroid nodule. The nodules are harder to palpate in children than in adults. When they are palpated, these nodules are significantly more likely to be malignant in children—approximately 25% of thyroid nodules in children are malignant compared with only 10% in adults.^{59,60} A thorough history, including details on previous radiation or benign thyroid disease and a physical examination, should be performed for all patients. Laboratory tests should include thyroid function studies. Imaging consists of initial thyroid ultrasonography and possible fine-needle aspiration biopsy. Further investigation with nuclear scintigraphy and CT may be performed.

Surgical excision is the first-line treatment of thyroid cancer in children. In cases of papillary and medullary carcinoma, a total thyroidectomy with parathyroid gland preservation is indicated with a low threshold to perform a selective neck dissection. For follicular carcinoma, a thyroid lobectomy may be performed. Radioactive iodine therapy is frequently used as adjuvant therapy in patients with papillary and follicular carcinoma, and adjuvant radiotherapy may be used in those with medullary carcinoma.

Although more than 65% of children have local lymph node metastases and 20% have distant metastases, the prognosis for children with thyroid cancer is good.⁶⁰ The average long-term survival rate for all subtypes is greater than 90%.^{61,62}

Nasopharyngeal Carcinoma

Nasopharyngeal carcinoma is a malignancy of the nasopharyngeal epithelium most often seen in adults. In children, it is usually found in adolescent males. The treatment is based on adult protocols, because this disease is rare in children. Surgical resection may be performed, although the disease may be unresectable as a result of the location and/or stage at presentation. Other therapy includes radiotherapy and chemotherapy (see Chapter 39 for further discussion of nasopharyngeal carcinoma).

Salivary Gland Carcinoma

There are three pairs of major salivary glands: the parotid, submandibular, and sublingual glands. In children salivary gland neoplasms account for less than 5% of these tumors overall. Salivary gland neoplasms are more likely to be malignant in children than in adults.⁶³ Benign neoplasms include pleiomorphic adenoma and Warthin tumor (papillary cystadenoma). Malignant lesions occur most frequently in the parotid gland (82%).⁶⁴ They include mucoepidermoid carcinoma, acinic cell carcinoma, adenocarcinoma, and adenoid cystic carcinoma, which collectively account for up to 90% of malignant salivary gland lesions in children.¹⁴

Pleiomorphic Adenoma

The most common benign neoplasm of the salivary glands is the *pleiomorphic adenoma*, which accounts for half of salivary gland tumors and more than 90% of benign salivary gland tumors in children.⁶⁴ Pleiomorphic adenomas present as slow-growing, painless, solid masses usually in the superficial parotid gland. They appear encapsulated on imaging but have microscopic extensions into the surrounding tissue, and thus superficial parotidectomy with facial nerve preservation is the treatment of choice. Warthin tumor, or papillary cystadenoma, is extremely rare in children.

Mucoepidermoid Carcinoma

Mucoepidermoid carcinoma is the most common malignant salivary gland lesion in children, representing more than half of malignant lesions in this population. It is primarily seen in adolescents. Most of these lesions in children are low-grade with a low rate of metastasis. Patients with mucoepidermoid carcinoma present with a slow-growing, painless, fixed mass in the parotid gland. With bulky disease, patients may have tenderness, dysphagia, trismus, and facial paralysis. Surgical excision is the treatment of choice. In parotid lesions this may be a superficial parotidectomy or total parotidectomy, possibly with neck dissection and/or radiation, depending on the tumor grade. The overall, 5-year survival rate is also affected by tumor grade. Low-grade lesions have more than a 92% 5-year survival rate, intermediate-grade lesions have a 62% to 92% 5-year survival rate, and high-grade lesions have a 0% to 43% 5-year survival rate.⁶⁵

Acinic Cell Carcinoma

Acinic cell carcinoma is the second most common salivary gland malignancy in children, making up 10% to 12% of these tumors. Like mucoepidermoid carcinoma, most of these lesions are low-grade and rarely metastasize. Presentation is also similar to mucoepidermoid carcinoma, with a painless, slow-growing mass, although these lesions are more likely to have associated symptoms of pain (30%) and facial paralysis (10%).⁶⁴ Parotid lesions are treated with parotidectomy (superficial or total) with facial nerve preservation. Adjuvant radiation is used with large tumors, positive margins, or recurrent lesions. Acinic cell carcinoma has a 10-year survival rate of more than 85%.⁶⁶

Melanoma

Melanoma is a cutaneous malignancy originating in the melanocytes—pigment-producing cells in the basal layer of the epidermis. It is relatively uncommon in children, accounting for 1% to 3% of pediatric malignancies.^{67,68} However, its incidence is increasing, and surgeons must keep this in mind when evaluating cutaneous lesions in children. The risk factors for melanoma include intermittent and intense ultraviolet light exposure, tendency to burn, freckly/fair skin, blue or green eyes, blonde or red hair, a family history of melanoma, and immunosuppression.⁶⁷ Other risk factors include skin conditions such as congenital melanocytic nevus, xeroderma pigmentosum, dysplastic nevus syndrome, atypical nevi, and acquired melanocytic nevi.⁶⁷

Pediatric melanoma can be categorized into four subgroups based on age⁶⁹:

1. Congenital (in utero to birth)
2. Infantile (birth to 1 year)
3. Childhood (1 year to puberty)
4. Adolescent (after puberty)

In congenital and infantile cases, melanoma may arise *de novo* in patients with a congenital melanocytic nevus or in utero from transplacental metastasis of maternal disease.⁵⁴ Giant congenital melanocytic nevi (diameter projected to be larger than 20 cm in adulthood) have been estimated to undergo malignant transformation in up to 12% of lesions.^{70,71} More recent data suggest that this is an overestimate, and that malignant transformation occurs in 0.7% to 2.9% of these lesions.⁷² Thirty percent of childhood melanomas are associated with a giant congenital melanocytic nevus. In adolescents the risk factors and profile of those affected are similar to those of adults, with the exception of a higher incidence in females.

In general, the presentation of children with melanoma is similar to that of adults with melanoma. Cutaneous lesions should be examined for signs of atypia. In children, as in adults, the trunk and extremities are the two sites most often involved (approximately 37%), followed by the head and neck region (29%).⁶⁸ Children are more likely to present with amelanotic lesions (Fig. 37-15). They may or may not present with palpable lymphadenopathy. The lesions may be difficult to distinguish clinically and histologically from a Spitz nevus, a benign lesion that appears so similar that they were once called *juvenile melanoma*.

A thorough history, including a family history, a physical examination, and a skin check are important in the diagnosis of melanoma. In part because it is so uncommon, many children have a delayed diagnosis or misdiagnosis (10%).⁷³ A high degree of suspicion and a low threshold to obtain a biopsy sample help to promptly diagnose lesions. A full-thickness biopsy for all suspicious lesions is essential to accurately assess thickness.

Surgical excision is the mainstay of treatment for melanoma. Excision margins are the same as those recommended for adult lesions by the National Comprehensive Cancer Network⁷⁴ (Table 37-4). A sentinel lymph node biopsy is performed in adults who have lesions greater than or equal to 1 mm and in those less than 1 mm with tumor ulceration, a high mitotic index, or other negative prognostic indicators. The role of a sentinel lymph node biopsy is not well described in children. However, it provides valuable information for disease staging and prognosis

and may direct further adjuvant therapy. The current recommendation is to perform a sentinel lymph node biopsy in children with the same indications as in adults.

The prognosis is affected by multiple factors. The stage at diagnosis, increasing thickness, a high mitotic index, ulceration or bleeding of the lesion, regional lymph node involvement, and systemic metastasis confer negative prognosis. In children, the 5-year melanoma-specific survival rate has been reported as 100% for in situ disease, 96.1% for localized disease, 77.2% for regional disease, and 57.3% for distant metastatic disease.^{68,73}

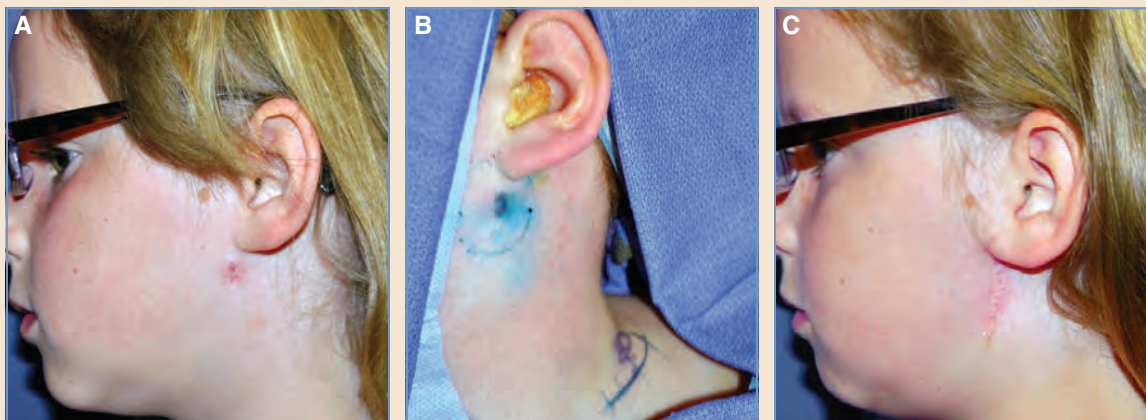


Fig. 37-15 A, Atypical amelanotic melanoma with spitzoid features in a 7-year-old girl. B, Surgical markings with a 1 cm margin. Methylene blue was injected for identification of the sentinel lymph node. C, The postoperative appearance after resection with negative surgical margins and a negative sentinel lymph node.

Table 37-4 Melanoma Resection Margin Guidelines of the National Comprehensive Cancer Network

Lesion Thickness (mm)	Recommended Margin (cm)
In situ	0.5
<1	1
1-2	1-2
>2	2

KEY POINTS

- Congenital and inflammatory causes account for most pediatric head and neck masses. In primary care settings, inflammatory causes are the most common reason for these masses, whereas congenital causes are more common in tertiary centers.
- Congenital lesions may not be diagnosed at birth. Diagnosis may be delayed until an inciting event—typically infection—causes the lesion to become more obvious.
- With improved prenatal ultrasonography, large, airway-compromising masses are increasingly detected, and an EXIT procedure may be planned at delivery.
- Cervical adenopathy is usually the result of infectious or inflammatory causes.
- Although malignant lesions of the head and neck are rare in children, if lymphadenopathy does not respond to initial treatment, the surgeon should consider malignancy in the differential diagnosis.
- Children treated for cancer have better outcomes for some malignancies when treated at comprehensive cancer centers.
- Pediatric plastic surgeons may be asked to obtain a biopsy sample of lesions of the head and neck, to excise lesions, and to perform reconstruction after lesion resection.
- Biopsies should be performed in a manner that does not compromise future reexcision or reconstruction.
- Surgery to excise lesions of the head and neck may require wide exposure and intraoperative nerve monitoring to adequately visualize and protect vital structures.

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38

Facial Paralysis and Facial Reanimation

Jeffrey R. Marcus • Gregory H. Borschel • Ronald M. Zuker



Cross-Facial Nerve Grafting for Smile: Extended and Standard; Segmental Gracilis Muscle Transfer: Extended and Standard; Microsurgical Facial Paralysis Reconstruction.



he term *facial paralysis* describes a physical inability to activate the muscles of facial expression. It is a condition of great variability. Impairment of the facial neuromotor system is associated with a wide range of causes. Even patients affected by conditions with similar causes can present in a variable manner and require different management strategies. Common to all affected by facial paralysis is concern over its effects—both functional and psychosocial. Clinicians immediately understand the functional implications of facial paralysis, including the consequences of oral incompetence and an inability to protect and lubricate the cornea. However, what makes facial paralysis such a unique medical entity is the human perceptual linkage between physical expression and emotion. During effective social interaction, emotion is associated with expression, which an observer perceives and reciprocates. This response is vital to effective social interaction. In a fraction of a second, we process visual information from a person's face and infer sex, age, status, mood, and intentions. The outcome of this processing influences our interaction with the owner of the face.¹ A disconnect in the sequence of expression and perception will compromise the quality of the interaction. Observers in general instinctively and acutely perceive asymmetry and associate it with unattractiveness. A person with unilateral facial paralysis presents an image discordant with an observer's expectations, potentially leading to incorrect assumptions regarding mood, personality, and intelligence.^{2,3} In *complete bilateral facial paralysis* (as in a child with Moebius syndrome), the facial display is symmetrical but unresponsive, leading to difficulties in conveying the emotional components of interpersonal communication. Regardless of the etiologic factors in unilateral or bilateral facial paralysis, coping with a relative inability to interact socially in this way is one of the most difficult aspects for patients with such physical constraints.

In this chapter we will discuss the approach to pediatric patients with facial paralysis. A practical etiologic classification of facial paralysis is presented, with emphasis on specific conditions. We review the normal and abnormal anatomy and the initial evaluation, pretreatment assessment, operative planning, surgical procedure, postoperative care, outcome analysis, and complications associated with the surgery.

EMBRYOLOGY

Motor neurons that organize to form the facial motor nuclei are derived from neuroblasts in the basal plates of the medulla and later shift cephalad to lie within the pons. Special visceral efferent fibers innervate striated skeletal muscles derived from the branchial arches. The primordial facial nerve is discernible in the fifth week of life and is composed of special visceral efferent neurons. The extratemporal facial nerve begins dividing into fascicles in the seventh week. Selected fascicles pass superficial to the parotid bud, which develops rapidly and ultimately surrounds the facial nerve. Branching of the facial nerve in this area is thought to occur as a result of fascicular separation from growth of the parotid gland and by expansive growth of the face.^{4,5}

The relationship between the development of the facial nerve and the external ear is important because of the concomitant time sequence. The external ear develops from the auricular hillocks, which arise from the first and second branchial arches. The hillocks begin to fuse by the end of the sixth week. Malformations of the first and second branchial arches, including the external ear (such as microtia), are thought to arise as a consequence of events between the sixth and twelfth weeks. Because this is also the critical developmental period for the facial nerve, auricular disorders should prompt investigation of facial nerve function. Such is the case in assessing a child with *hemifacial microsomia*, in which facial nerve dysfunction may accompany a constellation of first and second arch anomalies, including hemimandibular hypoplasia and microtia (Fig. 38-1).

A typical branchial arch contains an artery, a cartilaginous bar, a muscular component, and a nerve.⁵ The facial nerve is considered to be the nerve of the second branchial arch (Fig. 38-2). Myoblasts from the second branchial arch migrate from the neck to the head and give rise to the muscles of facial expression, the stapedius and stylohyoid muscles, and the posterior belly of the digastric muscle. In the absence of nerve supply, muscles will begin a developmental sequence and demonstrate an early spontaneous and stimuable response.⁶ However, without direct innervation, the muscle fibers degenerate and are resorbed, leaving a fibrous and sinewy residue.^{7,8} There are no distinct muscular planes. This has significant surgical implications, because identification of appropriate dissection levels becomes very difficult.

FACIAL NERVE ANATOMY

Central Nervous System

The facial nerve nucleus is located within the pons. Lesions in or distal to the pons will affect the entire ipsilateral facial musculature. These are the coordinated movements that provide eye protection, oral competence, and facial expression. The spontaneity component of facial expression requires input from the frontal lobe. Thus a true smile has connections to both the facial nerve nucleus and the frontal lobe, although the precise pathways have not been delineated.

The facial nerve fibers exit the facial nerve motor nucleus and pass dorsally encircling the nucleus of the abducens nerve and emerge at the lateral aspect of the pons in the angle formed by the junction of the cerebellum and pons (the cerebellopontine angle).⁹ The nerve then enters the internal auditory canal, proceeds through an infratemporal course, and exits the skull at the



Fig. 38-1 This 4-year-old boy has hemifacial microsomia. Associated anomalies include left anotia, mandibular hypoplasia, and unilateral facial paralysis involving buccal-zygomatic branches. A surgical plan must be presented to address the relevant anomalies in a cohesive time sequence.

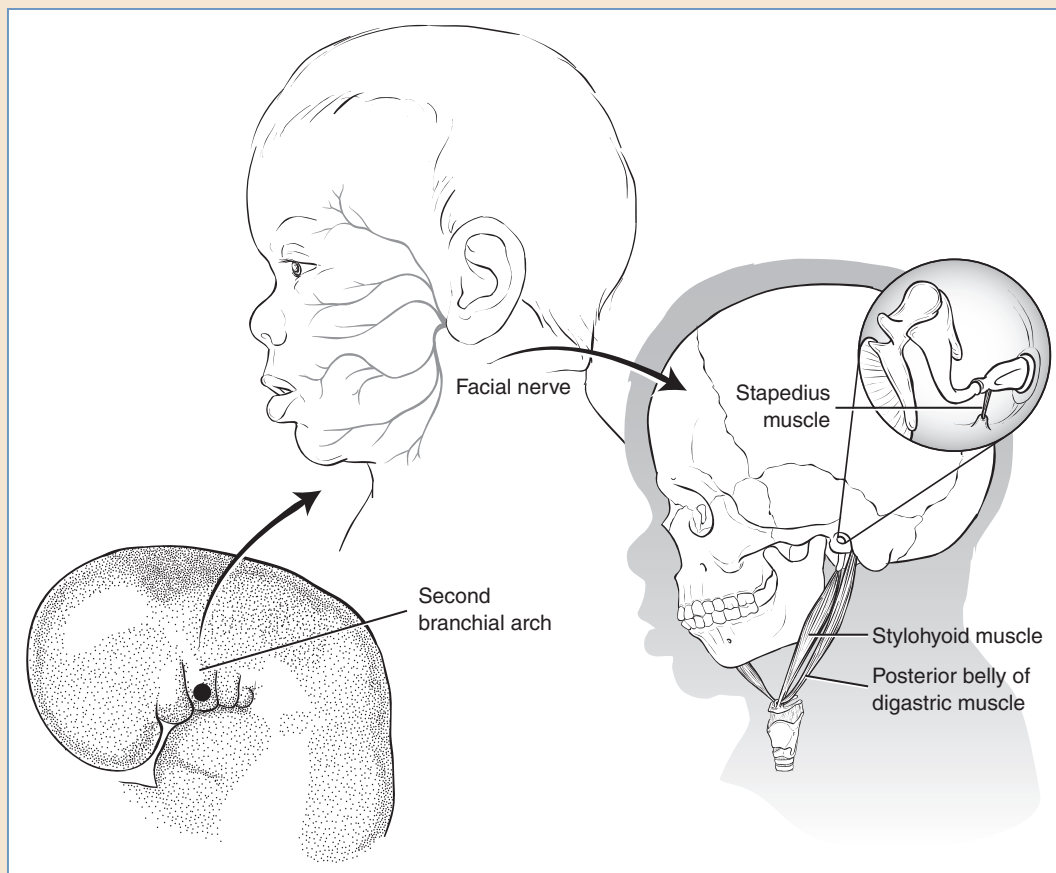


Fig. 38-2 The facial nerve is considered to be the nerve of the second branchial arch. It innervates the muscles of facial expression, the stapedius and stylohyoid muscles, and the posterior belly of the digastric muscle.

stylomastoid foramen. This infratemporal course of the facial nerve is the longest course of any nerve in the body through a bony canal (30 mm). It is divided into three segments: (1) labyrinthine (the narrowest site), (2) tympanic (horizontal), and (3) mastoid (vertical).^{4,10}

Extratemporal Course

The extratemporal course of the facial nerve is well characterized. Several key anatomic points merit discussion, along with associated references with which readers should be familiar. Baker and Conley^{11,12} and Davis¹³ described general variations of facial nerve anatomy. Classically, after initial branching to form the temporofacial and cervicofacial main branches, five terminal branches are designated: (1) temporal (frontal), (2) zygomatic, (3) buccal, (4) marginal mandibular, and (5) cervical. Significant crossover occurs among the zygomatic and buccal branches, causing variability of innervation to the central face. For this reason, traumatic injuries to distal branches (medial to the lateral canthus) in the cheek tend not to result in functional loss. This anatomic pattern is also relevant in reanimation of a paralyzed face, specifically with respect to innervation of the zygomaticus and levator muscles. Here, a specific distal branch can be sacrificed to provide a proximal source of contralateral (normal) innervation for grafting to the paralyzed side, without loss of innervation to the normal side. Such is not the case for the temporal, marginal mandibular, and cervical branches, which have little crossover, and transection of single branches can easily result in loss of function to the target musculature, such as the frontalis, the depressor labii inferioris, and the platysma muscles, respectively. Therefore considerable attention has been paid to the courses of these nerves in the context of rhytidectomy. Stuzin et al,¹⁴ Seckel,¹⁵ Dingman and Grabb,¹⁶ and Pitanguy and Ramos¹⁷ have published valuable articles on this subject. Branches of the facial nerve innervate target musculature on the deep surface consistently in all but three muscles: the levator anguli superioris, the buccinator, and the mentalis. These muscles are innervated on their superficial surface, a consequence of the deeper relative location of these muscles.

ETIOLOGIC CLASSIFICATION OF PEDIATRIC FACIAL PARALYSIS

Several authors have classified the extensive differential diagnosis of facial paralysis. May¹⁸⁻²⁰ noted more than 100 possible causes in a review of the medical literature between 1900 and 1996. In clinical practice, a systematic approach may be more useful rather than considering an inclusive list of specific diagnoses. Westin and Zuker²¹ proposed a classification system for facial paralysis intended to allow more general categorization and greater applicability to pediatric conditions. The major etiologic subheadings are indicated in Box 38-1. All presentations of facial paralysis may be appropriately assigned within this set of general categories, although the published classification system is far more detailed. Every case should be characterized not only by cause, but also by localization of findings (side-specificity and affected branches).

Pediatric cases are either congenital or acquired. The decisive factor is the likely causative event, rather than the age at presentation. Congenital conditions include any process or event occurring during prenatal development and are classified as syndromic or isolated nonsyndromic entities. Acquired conditions occur from the time of delivery through adulthood.

Congenital Conditions

Several diagnoses merit specific discussion. Unilateral facial paralysis present at birth may be the consequence of an obstetric injury; more often, it occurs as a developmental anomaly in isolated presentation or in conjunction with other physical findings.

Box 38-1 Etiologic Classification of Facial Paralysis**Congenital**

Syndromic
 Developmental
 Genetic
 Intrapartum
 Idiopathic
 Nonsyndromic
 Developmental
 Genetic
 Intrapartum
 Idiopathic

Acquired

Traumatic
 Intracranial
 Extracranial
 Tumor
 Benign
 Malignant
 Inflammatory
 Bacterial
 Viral
 Toxic
 Genetic predisposition

Neuromuscular disease
 Bacterial
 Viral
 Toxic
 Genetic predisposition

*Nearly all specific causes may be classified within this general outline. Further side and branch specificity and extent (complete versus incomplete) provide a comprehensive classification.



Fig. 38-3 This child with Moebius syndrome has bilateral complete facial nerve palsy and incomplete abducens and hypoglossal nerve palsies.

Hemifacial microsomia describes a heterogeneous group of conditions affecting the first and second branchial arch derivatives. The facial nerve is considered the nerve of the second branchial arch; as such, it may be affected in cases of hemifacial microsomia or Goldenhar syndrome, a specific related variant. The extent and severity of facial nerve involvement and branch specificity vary in hemifacial microsomia.

Bilateral facial paralysis among children is rare. The most common congenital presentation of bilateral facial paralysis occurs in Moebius syndrome, which is a congenital syndromic condition characterized by expressionless facies that remains unchanged through the entire range of emotions. It is seen in association with palsies of the sixth, ninth, tenth, and twelfth cranial nerves, the most common of which is abducens (sixth cranial nerve) palsy (Fig. 38-3). Moebius syndrome is frequently associated with limb anomalies, including club foot, syndactyly, and Poland anomaly. The etiologic factors in Moebius syndrome are unknown; however, in selected groups, associations to autosomal dominant inheritance have been proposed.²¹⁻²⁶

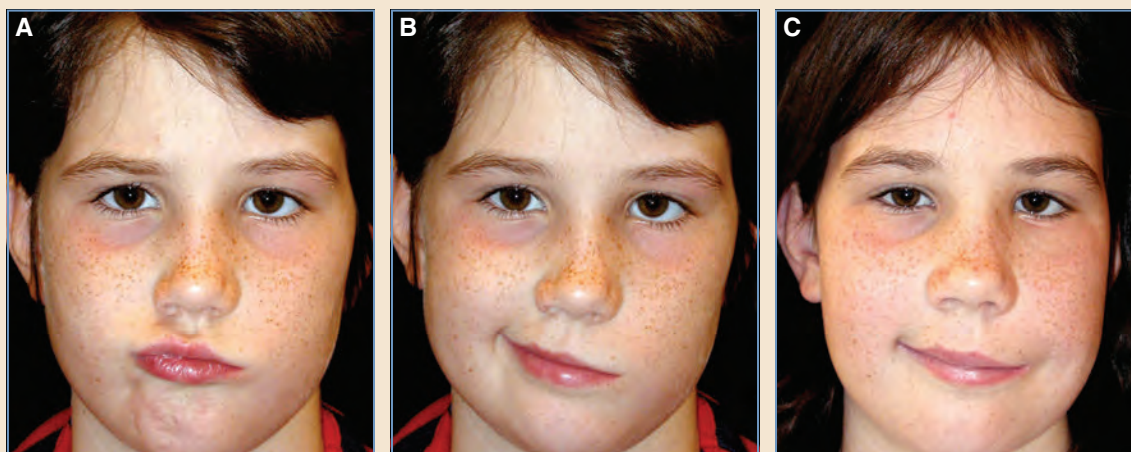


Fig. 38-4 This child presented with a posterior fossa medulloblastoma and underwent resection. **A** and **B**, Two years after surgery, she continued to have an incomplete seventh cranial nerve palsy primarily affecting buccal-zygomatic branches. She underwent cross-face nerve grafting and a free gracilis transfer. **C**, The patient is shown 6 months after reanimation.

Acquired Conditions

In the pediatric population, congenital anomalies are most prevalent. Acquired facial paralyses occur, although with a different distribution than in adults. In children, traumatic injuries and tumors are most commonly seen. Blunt, traumatic cranial base injuries may result in bilateral paralysis. Penetrating injuries, on the other hand, are generally isolated injuries to the extratemporal facial nerve branches. Tumors may be benign or malignant, occurring in an intracranial or extracranial site. Acoustic neuroma is the most common tumor-related cause of facial paralysis in adults. In children, ependymoma and medulloblastoma are more often seen (Fig. 38-4).

PREOPERATIVE ASSESSMENT

Patient History

Based on information from a patient's history, the surgeon should do the following: (1) establish the likely cause, (2) characterize the anatomy of the pattern of branch involvement, (3) determine functional status with relation to corneal protection and oral competence, and (4) chart the course of events, including onset, progression, and duration of the condition. Each of these priorities is necessary for planning the intervention.

The cause is a key factor in determining treatment and expected outcomes. A child with unilateral congenital facial paralysis would not be expected to regain function over time; in addition, the lack of native facial musculature would preclude the ability to reinnervate via cross-face nerve grafting or neurotization alone. On the other hand, in posttraumatic (acquired) facial paralysis resulting from blunt head injury, observation may be the most appropriate initial recommendation.

Determining the pattern of branch involvement and the presence of specific functional deficits is necessary for potential surgical planning. The first concern is protection of the cornea. Children with congenital facial paralysis (including Moebius syndrome) rarely have a complete

lack of corneal protection necessitating specific surgical procedures, such as gold weight insertion. Older children with acquired facial paralysis are more likely to have insufficient protective ability, more akin to that seen in adults overall.

The time course of events is necessary to determine the need and/or timing for intervention, particularly in patients with acquired deficits. Here, the status of the existing muscle motor endplates comes into question. For example, in the case of tumor resection with subsequent paralysis despite no known facial nerve or nucleus injury, understanding the length of time that has elapsed since the procedure helps surgeons to provide a prognosis and initial recommendations. A paralytic state that has persisted beyond 6 months without improvement is unlikely to resolve without intervention. In adults, reinnervation of existing musculature can be a reasonable option if up to 18 months of reinnervation can be provided from the onset of paralysis. Beyond this, the motor endplates tend to become less amenable to restoration of neural input. In pediatric patients, the specific time window may be greater.

Physical Examination and Grading

An examination should establish the historical factors mentioned previously. A detailed facial nerve examination is performed, with specific emphasis on the status of the cornea and the branch pattern and the extent of facial nerve involvement. Assistance and input from a trained facial rehabilitation specialist are important contributions. The extent of involvement in particular should be documented to facilitate recovery analysis and outcome measurement. Numerous grading systems are available for documenting the facial nerve status. A grading scale should be sufficiently sensitive to measure the natural course of a disability, from the onset of paralysis to the various states of recovery, and to detect changes over time or after treatment. It should have a high degree of reproducibility. The most commonly used grading scales include the House-Brackmann Grading Scale (HBGS) and the Toronto Facial Nerve Grading System (TFGS).²⁷⁻³⁴ The Facial Nerve Disorders Committee of the American Academy of Otolaryngology–Head and Neck Surgery adopted the HBGS as the standard for grading facial nerve recovery. The TFGS and the recently introduced MoReSS (movement, rest, secondary defect, and subjective) system³⁵ have addressed concerns regarding sensitivity and reproducibility with the HBGS. The practical yet comprehensive nature of these scales makes them suitable primary instruments for facial nerve grading.

Electrophysiologic Studies

The primary purpose of electrophysiologic studies in managing facial paralysis is to assist in determining the prognosis for recovery. It has limited use in congenital facial paralysis, because recovery is not possible in this group. In acquired facial paralysis, however, electrophysiologic studies may indicate the possibility of recovery; for these patients, surgical intervention is deferred. Nerve conduction studies and electromyography can be performed to follow facial nerve recovery. They are more helpful in adults recovering from Bell's palsy or resection of an intracranial tumor.

NONSURGICAL AND SURGICAL TREATMENT

In pediatric patients, surgical rather than nonsurgical management tends to be considered more often because of the predominant causes and their associated (often poor) prognoses. However, nonsurgical modalities may be beneficial in some cases, particularly those with incomplete paralysis. The distinction between complete and incomplete paralysis is sometimes unclear; it is based

on the presence of appreciable movement in the regional distribution of any facial nerve branch. Therefore a patient can have a complete paralysis in the distribution of the frontal branch, but an incomplete paralysis in the branches of the lower face. In *incomplete facial paralysis*, a theoretical potential for improvement exists, because some degree of neural input has sustained muscle fibers. A facial rehabilitation specialist may offer beneficial strategies using a variety of techniques. In children with complete or near-complete facial paralysis in any branch distribution, surgical treatment is the primary consideration. Surgical treatments are typically classified as static or dynamic procedures. *Static procedures* are intended to counteract paralytic effects through repositioning of structures and aiding in improved symmetry in repose. *Dynamic procedures* are intended to restore position and movement.

Nonsurgical Treatment

In unilateral incomplete facial paralysis, both sides of the face show function, with a disparity in the degree of function between the sides. A facial rehabilitation specialist may provide therapy to strengthen the weaker side and dampen the stronger side through exercise and biofeedback. This requires the understanding and cooperation of the patient. This can be a limiting factor in younger patients and/or those with developmental disability. Another nonsurgical adjunct in the treatment of facial paralysis is botulinum toxin (Botox). Usually, this is considered for marginal mandibular branch paralysis in which the normal side is overactive and causes considerable distortion, particularly with laughing and crying when the mouth is open. In this situation, the normal side can be injected with Botox to balance activity in the lower lip. A good test of effectiveness is an injection of bupivacaine, which paralyzes the musculature for 4 hours. The patient can assess the effectiveness and symptom resolution and then determine whether Botox would be beneficial. Although Botox injections have to be repeated every 3 to 6 months, long-term repetition may not be necessary, because some degree of adaptation may occur. If requested, a permanent surgical solution can be carried out later.³⁶⁻³⁹

Surgical Treatment

Static and dynamic operative strategies have been described for each of the regional territories of the facial nerve. The choice of procedures and their sequence must be individualized. Prioritization of functional deficits will guide the surgical itinerary. The treatment of the paralytic eyelid is generally the first consideration. Children have considerable skin elasticity; thus eyelid incompetence is not frequently seen. It usually can be controlled by drops alone in congenital cases. However, some acquired cases need surgery in childhood when control of corneal exposure is problematic.

Static Procedures

Treatment of the paralyzed upper and lower eyelid is indicated when dysfunction limits a patient's ability to protect the cornea, creating a risk for corneal exposure and sequelae. One of the most common means to provide corneal protection is through insertion of a gold weight implant to the upper lid.⁴⁰⁻⁴¹ The weight loads the tarsus and facilitates closure by gravity. Some refer to this as a static treatment, because it applies a constant force on the lid. Various weights are available depending on the severity of the condition and size of the patient. Selection of the appropriate implant and exact placement are crucial. Eyelid spring insertion is another option, which conceptually is more dynamic. However, it is technically more challenging.

Although gold weight implantation alone usually improves corneal protection, attention should also be given to the lower eyelids. Exposure results from an inability to close the lids, which is caused not only by loss of upper lid movement, but also by a descent in the position of the lower lid (lower lid ptosis). *Paralytic ectropion* is the most advanced stage of lower lid malposition; however, even mild malpositioning (manifested by degrees of scleral show) can sufficiently impair closure even with a properly placed upper lid weight. In general, lower lid procedures should be delayed until the degree of lower lid laxity is known. Therefore lid tightening procedures are generally not performed in the early stages after denervation (such as after a tumor ablation). In selected cases of more long-standing paralysis, gold weight implantation and lower lid tightening can be performed while the patient is under the same anesthetic. If this is chosen, the gold weight should be implanted before the lower lid procedure is performed so that the surgeon can estimate the amount of lower lid tightening required.

In mild-to-moderate conditions of lower lid laxity, canthopexy or canthoplasty can provide adequate support and repositioning of the lower lid. In severe lower lid ptosis and/or ectropion, a static tendon sling may be required. The sling is positioned along the lid margin, well supported at the medial and lateral canthi.³⁶ Brow malposition usually is not a major concern in pediatric patients until they have matured; over time, ptosis and relative differences in motion may become more apparent. Endoscopic brow lifting can be valuable for younger patients with unilateral brow paralysis.⁴² Standard cosmetic endoscopic brow-lift techniques are readily adaptable. The results appear to be durable for at least 1 year, with an acceptable aesthetic outcome. Recently, we have been using the Endotine absorbable anchor (Coapt Systems) for pediatric and adult patients, with acceptable results. The contralateral (nonparalyzed) side needs to be considered. In adults, if the contralateral side is ptotic, a bilateral brow lift may be needed. If the contralateral side is not ptotic (which is most often the case in children) and its activity results in obvious asymmetry, a frontal branch neurectomy, either surgically or with Botox, may be considered.

Long-standing cases of facial paralysis can lead to a shift or collapse of the nasal ala, resulting in relative nasal obstruction. Obstruction is caused by a medial shift in the alar base and by loss of dynamic ability to dilate the external valve. The cartilaginous structural support of the ala, provided by lower lateral cartilage, is generally not compromised. Therefore static procedures for nasal obstruction are designed primarily to reposition the ala. The alar base may be undermined through an upper buccal sulcus incision. The ala is repositioned and secured to the periosteum, or it may be secured using a small bone anchor. A tendon graft can also be used to reposition the nasal ala in severe cases with symptomatic obstruction.

In the lower face, the two primary concerns are loss of the ability to smile and impairment of oral competence. A wide variety of restorative procedures have been described for this region. Static procedures include the use of tendon/fascia, dermis, and synthetic materials placed to suspend the oral commissure.⁴³ For otherwise healthy pediatric patients, we do not generally recommend static procedures for treating the lower face. These patients are most amenable to dynamic reconstruction using microneurovascular muscle transplantation. Static suspension of the commissure may improve oral competence and rest symmetry but does nothing to restore the smile. The functional loss of smile is often the most concerning aspect of the condition to parents and children. Given the well-documented, successful experience in the young age group with microneurovascular muscle transfer and the associated lifelong benefits, it is our strong position to avoid the use of static procedures for the primary management of pediatric patients. For similar reasons, we do not recommend the use of regional muscle transfers, such as temporalis or masseter muscle transfers, in this group.⁴⁴

Dynamic Procedures

As opposed to static suspensory procedures, dynamic reconstruction provides movement, baseline muscle stimulation resulting in muscle tone, or both. A *dynamic procedure* requires a stimulatory source (a nerve) and an effector (a muscle). In pediatric patients, we think every effort should be made when possible to provide a motor source and an effector that most closely reproduces the normal state to provide coordinated, emotion-associated expression. As noted previously, this rationale excludes the use of regional muscle transfers such as temporalis or masseter muscle.

Several types of dynamic procedures have been reported. Their indications for reanimation can be explained according to the timing (and the mechanism) of functional loss. The timing situations that are encountered are acute, intermediate, and long-term or chronic. Acute and intermediate functional loss includes situations in which a stimulatory source is lost, but native musculature (that is, the effector) retains the ability to accept reinnervation based on existing, viable motor endplates. An example of an acute loss is a penetrating injury to the facial nerve along its course. The indication for repair/reconstruction is largely evident at the time of presentation. An intermediate presentation may occur because of the time elapsed from injury to referral or because of an inability to immediately establish the indication for surgery by nature of the mechanism and unpredictable recovery. An example is the onset of paralysis after a brainstem level tumor surgery in which the facial nerve was observed to be uninjured. In such circumstances, time must be allowed to establish the likelihood and potential outcome of nerve recovery. In acute and intermediate presentations, the strategy for reinnervation is based on reestablishment of the stimulatory source. The means by which to accomplish this may differ in the two settings. Patients in the long-term or chronic group, which includes those with congenital paralysis, have no native muscle effector present or capable of receiving stimulatory input and generating a response in the form of tone or movement. In this group, both the stimulus and the effector must be provided; therein lies the basis of free functional muscle transfer.

Nerve-based Dynamic Reconstruction: Nerve Repair, Nerve Graft, and Nerve Transfer

Where the facial musculature is potentially functional, surgeons can try to preserve the function of the facial musculature with a nerve-based reconstruction. The first form of nerve-based reconstruction for facial paralysis is a direct nerve repair. This can be done at any segment of the intracranial or extracranial path of the facial nerve and is always worthwhile. It may not provide perfect facial nerve function, but it can provide tone and baseline activity that can be very helpful for function. If the outcome of a nerve-based reconstruction is not adequate, an additional reconstruction can be performed.

Similarly, the second form of nerve-based reconstruction is a nerve graft to a defect in the facial nerve that cannot be directly coapted. This is very worthwhile at any point in the course of the facial nerve for the same reasons as noted previously, even though the result may not be perfect. Fig. 38-5 shows a patient with a nerve graft reconstruction with an additional microsurgical reconstruction. It demonstrates the tone and baseline activity of the nerve graft and the spontaneous excursion provided by the secondary microsurgical reconstruction.

The third form of nerve-based reconstruction that is gaining popularity reserves the function of the effector musculature with a nerve transfer. Various motor nerves, such as the hypoglossal, have been used as nerve transfers in an attempt to preserve function of the facial musculature. We prefer the motor nerve to the masseter, which can be fairly easily found and is described later.

In a nerve transfer scenario described by Klebuc,⁴⁵ the masseter nerve is transferred to the buccal branch of the facial nerve with a direct microcoaptation or with a small sural nerve jump



Fig. 38-5 **A**, This 7-year-old girl is shown preoperatively at rest. She had a facial paralysis secondary to cholesteotoma surgery carried out at 11 months of age. The damaged facial nerve was replaced with a greater auricular nerve graft and provided tone, eye protection, but little commissure elevation. **B**, When smiling, she had minimal commissure movement. **C**, She is shown postoperatively at rest after having a cross-face nerve graft–segmental gracilis muscle transplant as a dynamic reconstruction. This procedure is described in detail later in the chapter. **D** and **E**, Postoperative views show her small smile and full smile.

graft (Fig. 38-6). This patient had a complete facial paralysis, because the intratemporal component of his facial nerve had been destroyed in an acid burn. A nerve-based reconstruction was performed. The lower face was reinnervated with a nerve transfer from the motor nerve to masseter. The upper face was reinnervated using a cross-face nerve graft that connected the normal temporal branches of the facial nerve on the left side through an end-to-side graft, which then coursed across the forehead. The distal end of the graft was coapted end to end to the involved upper component of the facial nerve. Reinnervation occurred fairly rapidly to the lower face; activity was seen in about 6 months. Tone for the upper face and active movement did not begin until a year after the nerve grafting. However, the reconstruction provided tone in the upper face on the involved side and active movement with commissure elevation and nasolabial crease formation on the involved side.

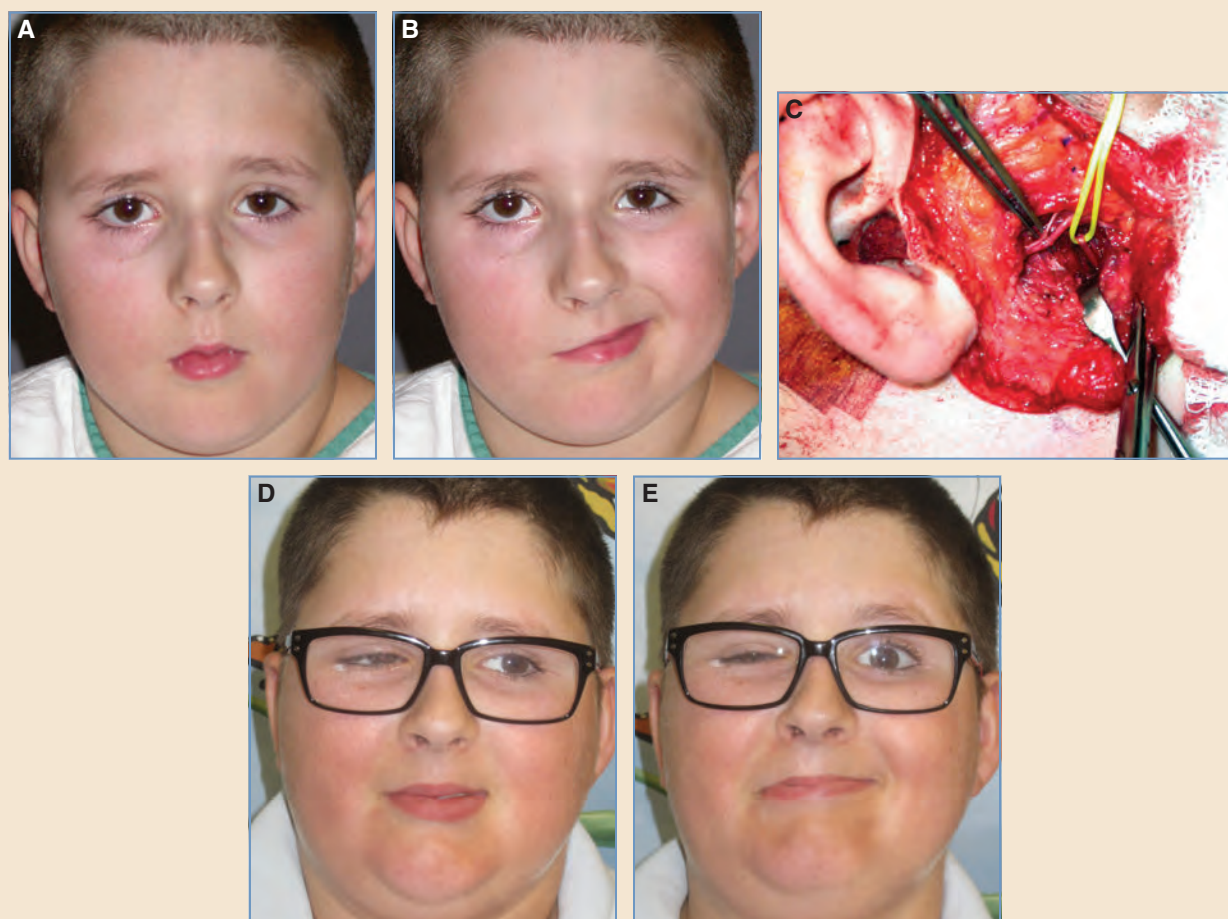


Fig. 38-6 A, This 10-year-old boy is shown preoperatively at rest. He had facial paralysis from an acid burn. B, His smile was asymmetrical, demonstrating an almost complete facial paralysis. C, Intraoperative view showing the motor nerve to the masseter in close proximity to the facial nerve in preparation for coaptation to the buccal branch of the facial nerve on the involved side. D, He is shown 2 years postoperatively at rest. Resting tone is improved. E, Smiling, he has a natural-appearing nasolabial crease from innate effector musculature.

One difficulty in using neurotization procedures intermediately is that the paralyzed branches at the time of surgery are “silent.” Their ability to respond to electrical stimulation of the facial nerve for the purpose of dissection/exploration diminishes significantly within days of injury. The *babysitter procedure* was developed in response to this problem.⁴⁶ It was originally described as a two-stage procedure in which movement is regained based on the babysitter. The hypoglossal is coapted end to end or end to side to the proximal extratemporal facial nerve. At the same setting, a cross-face nerve graft or grafts are coapted at the normal side, brought across to the paralyzed side, and left in proximity of the more distal branches, but they are not coapted. In the second stage, the distal branches are stimuable based on hypoglossal (babysitter) input; the appropriate branches are selected and coapted to the waiting cross-face grafts. This process addresses the “silent nerve” problem and allows selective grafting of the donor nerves at the second stage.

This concept is used in the second form of nerve transfer designed to preserve the intact facial musculature and was proposed by Marcus. In this scenario, the motor nerve to the masseter is coapted to the proximal part of the facial nerve—the part activating the lower face and the periorbital region. The masseter functions as a babysitter, coapted to the facial nerve in end-to-end fashion. Similar to Dr. Terzis' design,⁴⁶ simultaneously, two sural nerve grafts are coapted to selected normal side branches and brought across in proximity to distal branches but not coapted: one for the upper face and one for the lower face. Once facial movement based on the masseter transfer is demonstrated, the nerve is amenable to selective stimulation. In the second stage, the cross-face grafts are coapted end to end to selected distal branches of the paralyzed side. Distally, the facial nerve branches substantially; therefore the proximal connection of the masseter nerve is not disrupted, preserving the contribution of these fibers. With this approach, the facial musculature has strong innervations from the masseter (although little tone) and spontaneous, synchronous innervations with tone from the cross-face nerve grafts. Both of these nerve transfer techniques show great promise in the reconstruction of facial paralysis when the facial musculature is still able to be reinnervated.

Muscle-based Dynamic Reconstruction: Free Functional Muscle Transplantation

Functional muscle transplantation for facial reanimation has become the procedure of choice for longstanding or developmental pediatric facial paralysis. Techniques for microvascular facial reanimation have evolved considerably over the past 15 to 20 years. We will describe our own approach with this treatment. Many other variations are offered by other dedicated groups. Regardless of the approach, functional muscle transplantation for facial paralysis requires a source of neural input and an effector muscle. In unilateral facial paralysis, our approach is a two-stage sequence of procedures. In the first stage, neural input is established through a cross-face nerve graft from the contralateral (normal) side of the face. The second stage, free muscle transfer, is generally performed after 6 to 12 months to allow nerve growth through the graft.

Stage I: Cross-Face Nerve Graft We prefer to use the contralateral facial nerve for neural input, rather than direct neurotization or grafting to an ipsilateral cranial nerve (such as the hypoglossal). The advantage of the contralateral facial nerve is its potential to provide simultaneous, emotionally driven stimulus to both sides, allowing restoration of a spontaneous emotional smile. The surgeon identifies and uses the specific branch of the normal facial nerve that creates smile on the normal side, with the least amount of peripheral distracting movements. This branch should provide movement primarily to the zygomaticus major, the zygomaticus minor, and the levator labii. It should not significantly provide stimulation to the orbicularis oris or the nasalis. This is a crucial point and will be described in detail in the facial dissection section. Great care is taken to ensure sufficient facial nerve components to continue to innervate the normal side (see accompanying video entitled Cross-Facial Nerve Grafting for Smile: Extended and Standard).

The facial dissection begins with a preauricular incision and a submandibular extension on the normal side (Fig. 38-7, *A*). Dissection is carried anteriorly in the subcutaneous plane to the anterior border of the parotid. At this point, the dissection is carried deep, and the facial nerve branches exiting the parotid fascia are identified (Fig. 38-7, *B*). With the aid of the nerve stimulator, the specific branch to be used is selected. This is the specific branch that goes to the zygomaticus major, the zygomaticus minor, and the levator labii (Fig. 38-7, *C*). It should have minimal effect, if any, on the orbicularis oris. Additional branches that will innervate the facial musculature on the normal side are identified and preserved (Fig. 38-7, *D*). In this way, the surgeon can ensure that the division of the selected branch for cross-face nerve grafting will not weaken the normal side.

A tunnel is created from the site of the selected facial nerve through the subcutaneous fat into the upper lip. The tunnel courses across the sulcus in a preperiosteal plane to a site immediately overlying the maxillary canine root on the affected or paralyzed side. A small incision in the buccal sulcus is made to aid in the placement of the graft. The facial dissection is now complete. The sural nerve is harvested.

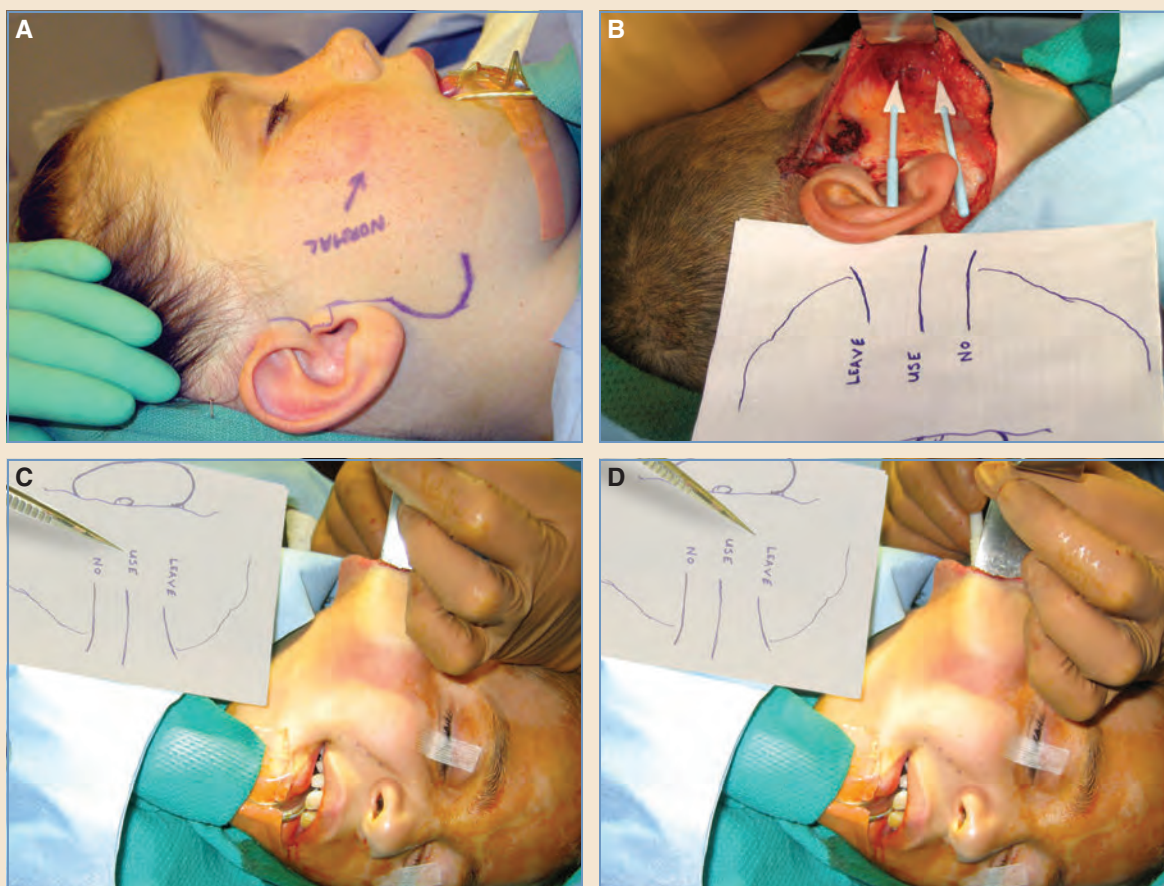


Fig. 38-7 **A**, A design for an extended rhytidectomy exposure on the normal side. The inferior extension of the incision, traveling in curvilinear fashion below the angle of the mandible, allows adequate exposure of the nerve. The same incision will be used on the contralateral, affected side in the second stage for free muscle transplantation. **B**, The facial nerve branches on the normal side are identified, and a facial nerve map is generated. The branch that will be used is identified as *use*. The branch that is not to be used is indicated as *no*. The branch that will continue to enervate the muscle supplied by the branch to be used is indicated as *leave*. **C**, The nerve branches are stimulated, and the specific branch that provides movement of the commissure consistent with a natural smile will be selected. This is the branch that will serve as the donor for sural nerve graft coaptation. The specific branches to the zygomaticus major and minor are identified with the aid of the nerve stimulator. It is helpful to use a nerve stimulator capable of generating sustained (tetanic) contractions to better differentiate muscle actions. This is the branch that will be divided and coapted to the cross-face nerve graft. **D**, Identification and preservation of another branch to the distal muscle targets that will be left to innervate these muscles after the selected branch is divided is imperative. This photo identifies the branch that will be left.

We prefer the sural nerve for cross-face nerve grafting. The entire nerve is not needed, and we prefer to use only the proximal part. The patient is placed supine with the hip flexed, abducted, and externally rotated, and the knee flexed at 90 degrees. A beanbag placed under the hip is helpful. Some centers place the patient in a prone or lateral decubitus position. The branch pattern of the sural nerve is somewhat variable, particularly in the lower third of the leg. The upper two thirds, traveling largely between the heads of the gastrocnemius, have fewer branches and a more suitable caliber; currently, we prefer this segment. A length of approximately 12 cm in a child older than 5 to 6 years of age can easily be harvested through a series of two or three stairstep incisions along the upper two thirds of the leg (Fig. 38-8, *A*). A 4 cm transverse incision is made approximately 4 cm distal to the popliteal fossa over the midportion of the proximal calf. This dissection is carried deep to the fascia, and very carefully the fascia is opened transversely. The sural nerve is identified between the two heads of the gastrocnemius muscle. Usually, it is quite superficial, but it may be buried between the heads of the muscle. It is dissected free and traced proximally to the popliteal fossa, where it is transected and brought out into the incision. At this point, a nerve stripper can be used to further dissect the nerve distally. The stripper is placed through the nerve and deep to the fascia and then gently advanced distally. It should clear about 12 cm of nerve. A second incision can be made in the midcalf, just above the distal end of the stripper. The distal portion of the nerve can be cut and the nerve removed for transplant (Fig. 38-8, *B*). The stripper is removed, and the incisions are closed.

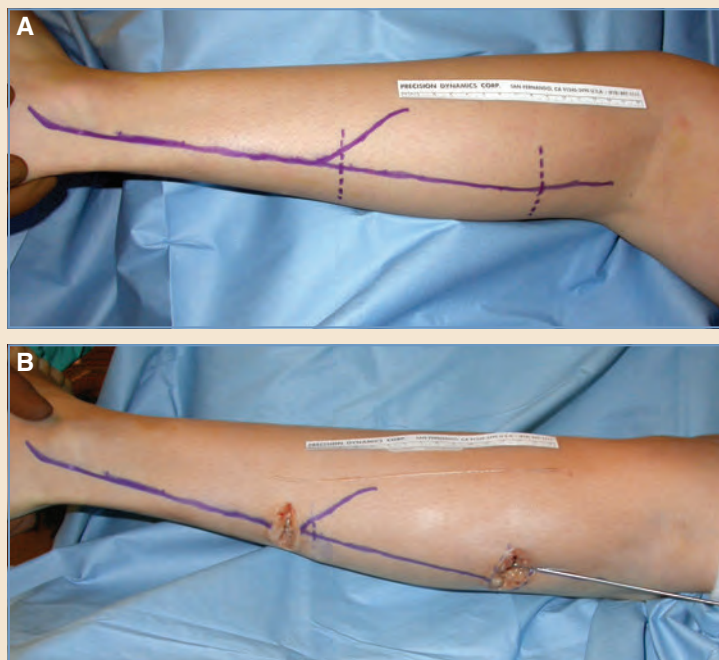


Fig. 38-8 *A*, The sural nerve is harvested from the proximal leg through transverse posterior-calf incisions. The nerve can be initially located in the proximal midline of the calf deep to the fascia and between the two heads of the gastrocnemius muscles. *B*, The nerve stripper has been passed from proximal to distal, a second incision made, the distal end of the nerve graft divided, and the graft removed. The stripper remains in place.

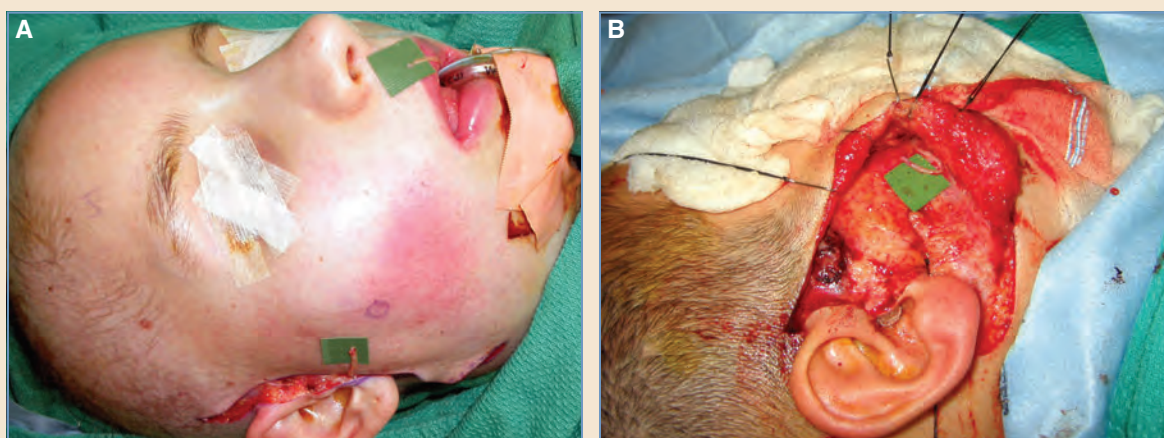


Fig. 38-9 A, Sural nerve graft in the face. One end is noted beneath the cheek flap; this will be coapted to the selected branch of the facial nerve. The other end is located in the buccal sulcus; this will be banked and will later serve as the neural input for the segmental gracilis muscle transplant. B, The micronerve coaptation has been carried out between the cross-face nerve graft and the selected branch of the facial nerve on the normal side.

If a stripper is not used, the proximal portion of the nerve is dissected as far distally as possible through the upper transverse incision. A second incision is made usually about 5 cm distal to the original incision. The nerve is identified, and the dissection is carried farther distally to the last stairstep incision, where the nerve is again identified, dissected distally, transected, and removed.

The sural nerve is then brought through the tunnel in the face (Fig. 38-9, A). One end is identified with a hemoclip and secured to the periosteum in the buccal sulcus with a nylon suture at the level of the canine. This will be helpful in identifying the nerve graft when stage 2—the muscle transplant—is carried out. The intraoral incision is closed. The other end is carefully coapted to the selected branch of the facial nerve on the normal side. The cheek flap is reflected anteriorly, and the microscope is brought in. A precise nerve coaptation is carried out between the sural nerve and the selected branch of the facial nerve under high-power magnification (Fig. 38-9, B).

A cross-face nerve graft procedure sets the stage for a muscle transplant. At the muscle transplant (6 to 12 months later), the buccal sulcus incision is reopened, and the nerve graft is identified. The clinical examination of the patient before surgery will provide evidence of nerve regeneration by the presence of a Tinel's sign (irritated nerve) over the maxillary canine. If the suitability of the nerve graft as a donor is in question, it may be helpful to send a sample to pathology for an intraoperative analysis. This will ensure the fascicles are healthy, viable, and satisfactory for reinnervating the muscle.

Stage II: Free Muscle Transfer Several muscles have been used for functioning muscle transplantation to the face for facial paralysis. These include the latissimus dorsi, pectoralis minor, extensor carpi radialis brevis, rectus abdominis, and gastrocnemius. However, we have found the gracilis to be the most useful. It has a consistent anatomy, can be harvested simultaneously with facial dissection, can be reduced in size to prevent bulk problems, has no functional loss when harvested, and leaves a relatively inconspicuous scar. The procedure is done with two teams (see accompanying video entitled Segmental Gracilis Muscle Transfer: Extended and Standard). Through a preauricular incision with a submandibular extension, a subcutaneous dissection plane is created. This extends anteriorly to the oral commissure and into the upper lip. Superiorly it



Fig. 38-10 **A**, A series of sutures is placed along the commissure and the upper lip. The placement of these sutures must be accurate to ensure proper creation of the nasolabial fold consistent with a natural smile. The sutures are placed in the sinewy material (which is representative of absent or atrophied facial musculature), with care to prevent inversion or eversion of the lip. **B**, The facial vessels are dissected. When divided, they can be transposed distally for microvascular anastomoses on the surface of the masseter muscle. **C**, The facial dissection in preparation for the muscle transplant is complete. The required length of muscle is the distance between the oral commissure (*short arrow*) and the root of the helix (*long arrow*); 1 cm is added to each end for suturing.

courses to the malar prominence and up over the temporalis fascia. Anchoring sutures for the muscle are placed in the oral commissure and upper lip. If the patient has a static droop of the oral commissure, then a suture is placed in the lateral part of the lower lip to provide support. One suture is placed in the oral commissure and, generally, two in the upper lip. The use of heavy (size 1 or 2) absorbable sutures helps to prevent the suture pulling through the muscle. With traction on these sutures, a well-defined nasolabial crease is created (Fig. 38-10, *A* through *C*). Just as the selection of the appropriate facial nerve branch is critical to the cross-face nerve graft, accurate placement of sutures is equally essential at this stage of the muscle transfer. The crease should be created at the same distance from the commissure as on the normal side. In the preoperative evaluation, the location of the crease and the vector of movement of the muscle are studied. The muscle is placed along the same vector seen on the normal side when the patient smiles. In cases of acquired paralysis, the surgeon visualizes the residual nonfunctioning orbicularis oris muscle and the insertion of the zygomaticus major and minor. This leads the surgeon to the proper plane for precise suture placement. If the sutures are placed too superficially, the lip will evert. If they are placed too deep, the lip will invert. Sutures are not necessary in the lower lip if little or no static droop is present in the oral commissure. They can be placed solely in the oral commissure and in the upper lip. In cases of congenital facial paralysis in which no native muscle is available to guide the surgeon, the sutures are placed in the sinewy material at the oral commissure and in the upper lip. This plane is difficult to identify, but the location of the facial artery is a very useful guide for appropriate suture placement.

The facial vessels are generally most suitable for revascularization of the muscle. The facial vein courses vertically upward just anterior to the anterior border of the masseter muscle. It is transected distally (superiorly) and reflected posteriorly for microvascular anastomoses. The facial artery courses anterior to the facial vein and toward the oral commissure. It is dissected to the commissure, transected, and reflected posteriorly. In this manner, the microvascular anastomoses are carried out above the surface of the masseter muscle, where visualization is straightforward (see Fig. 38-10, *C*).

Excess bulk is one of the most common problems after muscle transplantation to the face. It causes a facial asymmetry at rest but can be minimized in a number of ways. First, some of the excess subcutaneous fat in the cheek flap can be removed. The fat is removed in the area that will overlie the muscle transplant. Care is taken not to remove too much fat, because adherence to the dermis of the muscle will cause dimpling and distortion. Thus only the deep fat component is removed for contour purposes. We have found removal of part or all of the buccal fat pad to be extremely helpful in reducing bulkiness and excessive fullness in the cheek. This can be accomplished by spreading the muscle in the preauricular and temporal area at the time of transplantation so that it is not as thick as it might be just below and posterior to the malar prominence. Perhaps the most important factors are that the gracilis muscle can be longitudinally divided, and only 33% to 50% of the circumference of the muscle needs to be used, as described in the following discussion.

After the plane of dissection in the cheek has been prepared, the required length of muscle is determined. This is the distance from the oral commissure to the root of the helix when the oral commissure is positioned at the same level as the unaffected side. With slight traction on the sutures, the surgeon can position the oral commissure appropriately and then measure the distance from the commissure to the root of the helix. This is the functioning length of muscle that will be necessary. Generally, we harvest one additional centimeter on either end to accommodate suturing. Thus the functioning length plus 2 cm is the length of the gracilis muscle that will be required for reconstruction.

Muscle Elevation An upper medial thigh incision is made in a longitudinal fashion. A line is first drawn from the origin of the gracilis to its insertion on the tibia. The actual incision will lie 1 cm posterior to this line so that it will be less visible when viewed from an anterior perspective. The incision measures approximately 10 cm and is positioned such that the pedicle will lie centrally along the incision (Fig. 38-11). The dissection is carried through the subcutaneous fat to and through the fascia overlying the gracilis. A myocutaneous perforator coming from the gracilis muscle belly is often seen. This can aid the surgeon in identifying the pedicle, which should lie at the same longitudinal level as the perforator. The pedicle is identified and dissected to its origin. The vessels course at a 90-degree angle from the muscle, and the nerve courses at a 45-degree angle toward its takeoff from the obturator nerve just distal to the obturator foramen. The muscle is dissected circumferentially, and the neurovascular pedicle is dissected back to its origins. The muscle is inspected for size reduction. Generally, the pedicle comes in on the deep surface of the anterior part of the muscle. Only 33% to 50% of the circumference of the muscle is required, but this will vary from patient to patient, depending on the size of the muscle (Fig. 38-12, *A*). Occasionally, the pedicle comes into the central part of the muscle. In this situation, segments from both the posterior part and the anterior part could be removed to provide the appropriate bulk reduction. The segment to be harvested is then marked. Generally, the pedicle is placed in the midportion of the muscle, but sometimes it can be altered to accommodate the position of the motor nerve and reduce the time of reinnervation.

In a unilateral facial paralysis situation, the pedicle should be closer to the proximal part of the muscle. This is the part that will be placed at the commissure and upper lip. Thus the motor nerve to the gracilis will course from the cheek into the upper lip through a tunnel for neural coaptation in the upper buccal sulcus. The muscle is cut to the appropriate length (Fig. 38-12, *B* and *C*). Mattress sutures are placed along the edge of the muscle that will be inserted to the oral commissure and upper lip. These mattress sutures are placed close to the edge of the muscle and throughout its length. They will be used to anchor the muscle with the sutures that have been

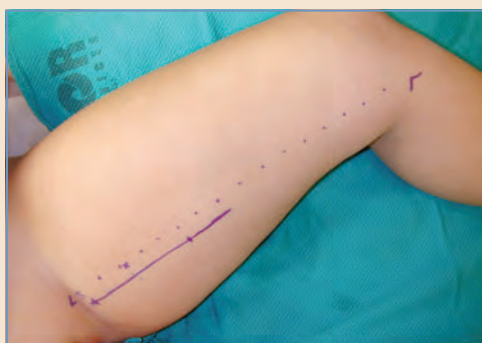


Fig. 38-11 The incision for the segmental gracilis harvest is positioned so that the pedicle will lie at about the midpoint of the incision. This allows the surgeon flexibility in designing the muscle flap with appropriate length on either side of the pedicle.

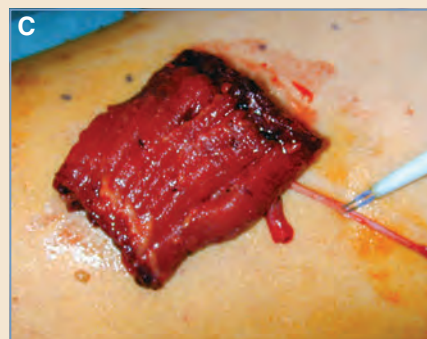


Fig. 38-12 A, The gracilis muscle may be split longitudinally to provide an innervated segment of suitable dimensions while preventing unnecessary bulk. B, The length of muscle needed to maintain the rest position of the commissure even with the normal side is determined. The length of muscle needed on either side of the pedicle is measured to place the pedicle at the appropriate position for anastomosis to the donor vessels. C, The muscle, ready for insertion into the face, is stimulated with a nerve stimulator.

placed in the oral commissure and upper lip. They will prevent sliding of these sutures through the muscle and malpositioning of the muscle. Should this occur, it is almost impossible to effectively correct.

Muscle Transplantation The muscle is transplanted to the face. The anchoring sutures are secured behind the mattress sutures. An additional bite into the tissues of the oral commissure and upper lip is placed, and the suture is placed once again behind the adjacent mattress suture (Fig. 38-13, A). Next, the muscle is slipped into position and secured. In this way, the muscle is not only appropriately positioned to create a natural nasolabial crease, but also is secure in this position, preventing suture pull-through. The motor nerve to the gracilis muscle transplant is placed through the previously created tunnel into the upper lip. Adequate length should be available for a tension-free nerve repair. The muscle is stretched temporarily to facilitate the microvascular anastomoses. Great care is taken not to kink the vessels of the pedicle or twist the facial artery or facial vein. Typically, the facial vein is repaired first and the facial artery second. After this, the motor nerve is repaired. After the nerve repair is completed, the vessels are inspected for patency. The entire muscle should be well vascularized.

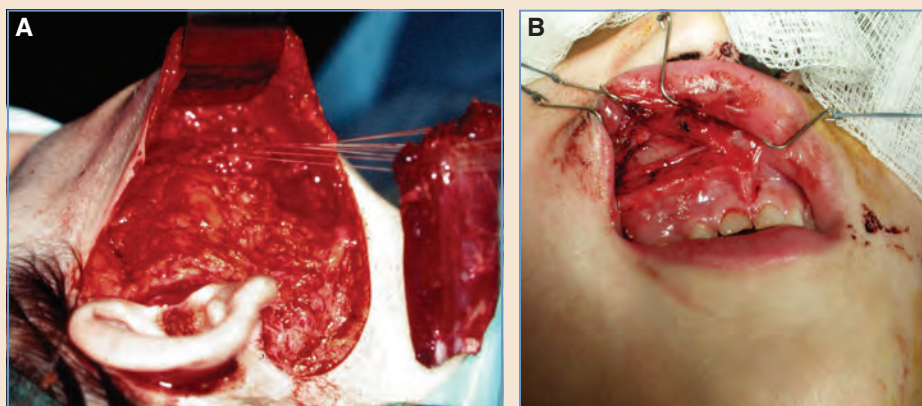


Fig. 38-13 **A**, The segmental gracilis muscle is carefully inserted into the facial pocket. **B**, Unilateral facial paralysis. Repair of the nerve to the gracilis muscle and a cross-face nerve graft in the buccal sulcus, which is done using an operating microscope.



Fig. 38-14 The muscle is secured to the oral commissure and upper lip. The origin of the muscle is inset along the temporal fascia after the vascular anastomoses have been completed. Ideally, the tendinous origin will be available to secure the muscle at this inset.

The head is turned, the operating microscope shifted, and appropriate exposure established for the nerve coaptation in the upper buccal sulcus (Fig. 38-13, *B*). The motor nerve to the gracilis and the sural nerve graft are generally an excellent size match. After the epineural nerve repair under no tension, the buccal sulcus incision is closed.

The next important step is to secure the origin of the muscle. The muscle is gently stretched to the temporal fascia. It should course just below the malar prominence to prevent excess bulk in this region. It is spread over the temporal fascia and to the area just below the anterior helical margin in front of the tragus. The muscle is secured with mattress sutures to the temporal fascia using heavy 1 suture. The amount of tension should allow hardly any movement at the oral commissure (Fig. 38-14). The oral commissure should be in a neutral position to the opposite side to prevent excess tension on the muscle and distortion at rest. This will maximize the muscle's excursion with contraction. A silicone drain exits through a stab wound behind the ear. The cheek flap is repositioned without tension on the overlying muscle transplant.

Bilateral Facial Paralysis

The approach is altered when an ipsilateral facial nerve and contralateral facial nerve are available. This is most frequently encountered in Moebius syndrome; it is also seen in patients who have had brainstem tumors removed and have a residual bilateral seventh nerve paralysis. We initially used the hypoglossal nerve in such situations to innervate muscle transplants but have not found it to be satisfactory. Power is not sufficient to effect adequate excursion, and the tongue may have an unacceptable motor dysfunction. Children with Moebius syndrome often have other speech difficulties, and reduction in tongue mobility could further impair speech. Although end-to-side repairs to the hypoglossal nerve have been described, they do not provide sufficient innervation to the muscle for effective contraction. We prefer to innervate the muscle in such cases with the motor nerve to the masseter muscle.

The motor nerve to the masseter muscle (that is, the masseteric nerve) provides a robust source of donor motor axons for use in rehabilitating facial paralysis. It can be used as a nerve transfer directly into an acutely injured facial nerve,^{45,47} as described previously; it can also be used to innervate a muscle transplant, such as the gracilis muscle.

Surgeons may wish to use the following landmarks as a guide to facilitate the dissection of the motor nerve to the masseter, as described by Borschel et al.⁴⁸ First, the masseteric nerve is located 3 cm medial to the tragus and 1 cm inferior (3 and 1) to the zygomatic arch.

The masseter muscle consists of three lobes: superficial, intermediate, and deep. The motor nerve to the masseter travels inferomedially within the space between the intermediate and deep lobes.⁴⁹ Electrocautery is used to make a transverse incision at the 3-and-1 point in cases of complete facial paralysis. In cases in which some facial nerve functions remain intact, this initial portion of the dissection is carried out using blunt dissection. On entering the fascia, the masseter muscle is readily identified, and dissection proceeds with blunt scissors dissection and placement of Ragnell retractors. Every time the scissors are opened to dissect, the Ragnell retractors are replaced, in sequence, to the level of the deep lobe of the masseter muscle. Because it can sometimes be difficult to know when the deep lobe has been reached, probing the masseter muscle with an electrical stimulator/nerve locator (such as a battery-powered stimulator made by Medtronic) is quite useful. Masseter muscle activity is usually visualized using these maneuvers, leading the surgeon to the masseteric nerve (Fig. 38-15). Once identified, the masseteric nerve is dissected from its bed. Usually, a region of about 1 cm contains few branches. The nerve is transected and delivered out of the masseter muscle for coaptation.⁴⁸



Fig. 38-15 The approach to the motor nerve (arrow) to the masseter muscle.

The motor nerve to the masseter muscle generally has one or two large fascicles and provides excellent power to the transplanted muscle.^{47,50} When innervation is derived through the masseteric nerve (ipsilateral), the motor nerve to the gracilis may be shortened—an advantage for quicker and more effective reinnervation of the muscle. The nerve is coapted under high magnification. The nerve repair is completed before the muscle origin is secured, because it will lie beneath the final resting position of the transplanted gracilis muscle. The muscle is appropriately positioned. The wound is thoroughly irrigated and the cheek flap closed again over a silicone drain exiting through a postauricular stab wound.

POSTOPERATIVE CARE

An optimal environment for muscle revascularization is critical. Thus adequate fluid resuscitation, appropriate pain control, and protection of the operative site are essential. Patients are generally maintained on bed rest for the first day and then are gradually mobilized. Parents are instructed to avoid any significant contact with the operated site for 6 weeks. In our practice, perioperative antibiotics are given, but anticoagulants are not. Patients are instructed to not consume caffeine for 6 weeks to minimize the chance of vasoconstriction. When the motor nerve to the masseter



Fig. 38-16 Unilateral facial paralysis. A and B, This patient is shown preoperatively at rest and smiling. C and D, Approximately 5 years postoperatively, she is shown at rest and smiling.

is used (or in rare cases the ipsilateral facial nerve), innervations occurs in about 8 to 10 weeks, because the distance between the nerve coaptation and the muscle is minimized. When a cross-face nerve graft is used, the innervation may take up to 5 to 6 months.

Rehabilitation can be very helpful to improve the muscle excursion and symmetry of smile. Once the muscle has been reinnervated, a series of fairly simple exercises can be initiated. This can strengthen the transplanted muscle and improve excursion. With the use of biofeedback, symmetry of movement can be improved. This is done by performing the exercises in front of a mirror, where the amount of movement and the extent of excursion can be assessed by the patient. This should be done two to three times a day for 4 to 5 minutes. The patient will gain an understanding of what needs to be done with his or her newly transplanted muscle to achieve the symmetry of movement and the spontaneity desired. In unilateral facial paralysis treated with an appropriate cross-face nerve graft, spontaneity is not a problem; however, the extent of excursion may be problematic. Strengthening exercises can be of considerable benefit (Fig. 38-16). When the motor nerve to the masseter muscle has been used, complete spontaneity may not be obtained. However, a degree of spontaneity is possible with exercises performed in front of a mirror using biofeedback. This rather simple rehabilitation process should be carried out for about 6 months (Fig. 38-17).



Fig. 38-17 Moebius syndrome with bilateral facial paralysis. **A** and **B**, This patient is shown preoperatively at rest and smiling. **C** and **D**, Twelve months postoperatively, he is shown at rest and smiling.

RESULTS

The results of muscle transplantation for facial paralysis can be quite dramatic. However, to date, we are not able to perfectly replicate “normal” appearance and function. The muscle insertion can be placed close to the oral commissure and will achieve reasonable symmetry, but the nasolabial crease is very difficult to create in its superior portion (Fig. 38-18). Some patients show the lower teeth on the unaffected side when they smile. This is very difficult to duplicate with a single-vector reconstruction. Commissure excursion has improved significantly over the years as a result of optimizing muscle excursion with better, long-lasting fixation, appropriate tension, and increased fiber length. It is also improved as a result of greater motor input with the use of larger segments of the facial nerve from either the ipsilateral side or the contralateral side. We think that the use of a short-nerve graft with no branching has been helpful in providing additional input into the transplanted muscle. Nevertheless, muscle excursion often does not match the unaffected side and remains a problem, particularly with cross-face nerve graft–muscle transplantation combinations in older patients.

Symmetry is a concern in bilateral cases. Despite considerable effort directed at creating symmetrical insertions and symmetrical vectors, the result is not always perfect (Fig. 38-19). However, an excellent smile can be restored in most bilateral cases, particularly in patients who had no movement previously. The excursion that can be created with the motor nerve to the masseter is impressive. It approaches the normal excursion of the oral commissure on the unaffected side.

COMPLICATIONS

The complications of muscle transplantation to the face for facial paralysis can be described as *early* or *late*. The early complications are bleeding and infection. Even with careful hemostasis, hematomas can develop. This is particularly true on emergence from anesthesia, with increased blood pressure. Although we generally give perioperative antibiotics to prevent infections, they can occur. We found that thorough irrigation at several points during the facial dissection can be effective in reducing the incidence of infection. Abscess formation is very troublesome; it often occurs directly beneath the muscle and surrounding the vascular repairs. An open approach to abscess evacuation is necessary. We have found that the muscle remains viable, and the vascular repairs are relatively resilient. Muscle function can be restored even after significant abscess evacuation.

Late complications are most concerning and very difficult to correct. The most common is excess bulk at the site of the muscle transplant. Prevention of this complication is ideal. Treatment involves an open approach with removal of some of the subcutaneous fat and some of the muscle bulk in a tangential fashion. A second complication is disruption of the muscle insertion from the oral commissure and upper lip. This is an extremely troublesome complication and very difficult to correct. It is sometimes possible through an open approach to release the muscle from the surrounding scar tissue and to reposition it to the oral commissure and upper lip. However, great care is required to prevent excess tension and distortion of the lip. The scar tissue should be released and the muscle stretched to its original length. Successful repositioning is not always possible. A third complication involves excessive initial tightening of the muscle and resultant pulling of the oral commissure at rest. This can be improved by releasing the origin of the muscle in the preauricular region, allowing it to slide anteriorly. The muscle should be freed very carefully on its undersurface, where the motor nerve is located.

Another complication that is difficult to correct is inadequate movement. This may be the result of poor neural input or reduced viability of the muscle; however, the actual cause is often obscure. Occasionally, muscle strengthening exercises and rehabilitation are helpful in gaining

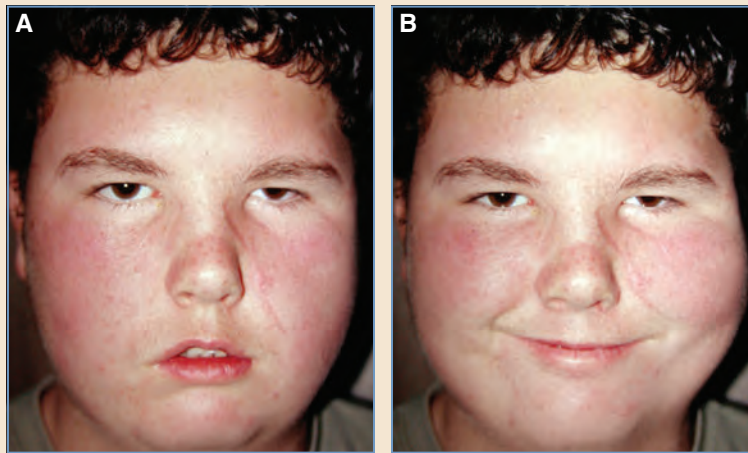


Fig. 38-18 Facial paralysis. **A**, This patient is shown postoperatively at rest. **B**, He is smiling 18 months postoperatively. He has incomplete formation of the upper component of this nasolabial crease.



Fig. 38-19 Bilateral facial paralysis of Moebius syndrome. **A-C**, This patient is shown preoperatively at rest, with maximal facial animation, and with maximal eye closure. **D-F**, She is shown postoperatively at rest, with a small smile (with fairly symmetrical nasolabial crease formation), and with a full smile (with asymmetrical nasolabial crease formation).

further excursion. We have also seen success with the use of electrical stimulation therapy. We have observed an increase in electrical activity and power over the course of 2 to 3 months of therapy. Electrical stimulation has been discouraged in the past in facial paralysis patients. However, the criticisms have focused primarily in adult Bell's palsy patients. In this group, stimulation of the native facial nerve can exacerbate synkinesis. In free muscle transplant patients, synkinesis is not a problem, because stimulation of the nerve graft is only directed to the transplanted muscle. If this is ineffective, the only option for improvement is to repeat the muscle transplant. Surgeons may wish to use a more powerful nerve. If the first reconstruction involved a cross-face nerve graft, a second muscle transplant might do better with innervation through the very powerful motor nerve to the masseter muscle.

CONCLUSION

Muscle transplantation for facial paralysis has provided an excellent means of dynamic restoration of facial animation. For near-normal spontaneity, the contralateral facial nerve can be used in a cross-face nerve graft. Great care is needed to select the appropriate branch. For a muscle transplant, we prefer the gracilis muscle. Accurate muscle placement and muscle fixation are necessary for the procedure to be successful. It is imperative to reduce bulk using only a fraction of the transplanted muscle and to remove the overlying fat and the buccal fat pad. Last, the muscle must be under the appropriate tension so that it barely moves the oral commissure and upper lip. In general, for unilateral facial paralysis, a two-stage approach is used. An initial cross-face sural nerve graft is carried out; 6 to 12 months later a free muscle transfer is performed. A key technical factor is the selection of the appropriate branch of the contralateral facial nerve in the first stage. In patients in whom neither the ipsilateral nor the contralateral seventh nerve is available, the use of the motor nerve to the masseter muscle to innervate the transplant has proved most effective.

KEY POINTS

- Facial paralysis can impose a major psychosocial barrier. A disconnect in the sequence of expression and perception compromises the quality of the interaction.
- Facial paralysis reconstruction can be either static or dynamic. Because of skin elasticity, the eyelids only rarely need surgery in children. In older adolescents and adults, a canthoplasty may be required. However, the lower face in children does best with a dynamic reconstruction rather than static sling support.
- The most critical aspect in a cross-face nerve graft procedure is the selection of the correct branch of the facial nerve on the normal side. Another branch needs to remain intact to innervate the musculature of the normal side. Harnessing sufficient axonal input is essential for adequate and effective muscle function.

- The most critical points in a muscle transplant procedure are (1) to correctly place the anchoring sutures in the commissure and upper lip, (2) to secure the muscle so it does not slip away from its insertion site, (3) to prevent excess bulk by using only 50% or less of the gracilis muscle circumference—removing the buccal fat pad and some subcutaneous fat in the area under which the muscle will lie—and (4) to use an effective motor such as the motor nerve to the masseter muscle.
- Late complications include excess bulk, muscle malposition, muscle tightness with distortion, and inadequate movement. These are extremely difficult if not impossible to correct secondarily. Thus surgeons should be aware of them and try to prevent them at the muscle transplantation procedure.
- Facial paralysis reconstruction in children can be extremely rewarding. However, each stage is fraught with potential pitfalls and complications that may not be correctable. Great care in patient selection and attention to detail in surgical execution are advised.

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39

Pediatric Cranial Base Masses

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he cranial base is composed of the paired frontal, paired temporal, ethmoid, sphenoid, and occipital bones and forms the floor of the cranial cavity. It also separates the cranial cavity from the facial structures, including the sinuses and nasal and oral cavities. Cranial base lesions are relatively uncommon in children, and the lesions that do occur are more likely benign than those in adults. A greater percentage of benign lesions and increased rates of complete tumor resection resulting from defined tissue planes in children portend a better prognosis than in the adult population.¹ However, there are specific challenges when one is diagnosing and treating these lesions in children. The ability to surgically approach skull base lesions has long been one obstacle to their treatment; this difficulty is compounded by the small size of the pediatric cranium. The differing anatomic proportions and the presence of growth centers in the developing craniofacial skeleton pose additional hurdles in planning adequate exposure for resection.

PRESENTATION

Cranial base lesions, whether in children or adults, may be asymptomatic or can present with various symptoms, depending on the size and location of the tumor. Common complaints include vague symptoms such as headache, confusion, and nausea and vomiting. Symptoms may also be indicative of tumor location, including specific cranial nerve palsy, epistaxis, anosmia, difficulty breathing, hearing loss, tinnitus, vertigo, nausea, diplopia, and proptosis. Especially in younger children, it may be difficult to elicit specific symptoms before a lesion is sufficiently large to produce symptoms resulting from mass effect.

Box 39-1 Lesions of the Anterior Fossa**Congenital Lesions**

- Encephalocele
- Nasal dermoid
- Glioma

Benign Lesions

- Fibrous dysplasia
- Juvenile nasopharyngeal angiofibroma

Malignant Lesions

- Esthesioneuroblastoma
- Nasopharyngeal carcinoma

DIAGNOSIS

Patients may be seen by a variety of specialists before a definitive diagnosis is reached; this may include plastic surgeons, neurosurgeons, and otolaryngologists, based on the perceived issue and chief complaint. Skull base tumors should be considered part of the differential diagnosis, since they may present with a variety of seemingly unrelated symptoms. A thorough history and physical examination are essential to diagnosis. In particular, the onset and duration of symptoms and exacerbating factors should be noted. Depending on the age of the child, much of this information may come from parents or other caregivers. In addition to history and physical examination, imaging is critical to diagnosis and treatment planning. CT and MRI are the primary imaging modalities used when a skull base lesion is suspected. CT provides greater bony detail and illustration of sinus disease, whereas MRI provides greater soft tissue detail and is helpful for evaluating the brain parenchyma and meninges. The two modalities are frequently used in a complementary fashion.^{2,3} CT scans in particular should be performed judiciously, with described dose-reduction strategies, especially in younger children.⁴ Angiography remains the benchmark for diagnosis of suspected vascular lesions. Although efforts must be made to eliminate unnecessary radiation exposure in children, as many as 37% of these lesions may have some intracranial involvement that necessitates radiographic imaging.⁵

Tumors of the skull base may be divided anatomically into those involving the anterior, middle, or posterior fossa. Tumors of the anterior fossa are most likely to present to the plastic surgeon, given their location and propensity to present with facial signs and symptoms. These lesions are also the most likely to require plastic surgical expertise with craniofacial surgical approaches. However, regardless of who these patients see, a multidisciplinary team approach is quickly becoming the standard for treatment. Plastic surgeons are often called on to assist with a surgical approach to the cranial base because of their expertise with soft tissue management and experience with complex osteotomies.

LESIONS IN THE ANTERIOR FOSSA

Cranial base lesions in children are rare, and in particular lesions of the anterior fossa are much less common than those in adults. Lesions in this region may be characterized by their natural history as congenital, benign, or malignant (Box 39-1). Surgical planning for resection of these

Table 39-1 Encephalocele Classification Based on Anatomic Location

Anterior/sincipital	Nasofrontal Nasoethmoidal Nasoorbital
Basal	Sphenoorbital Sphenomaxillary Sphenoethmoidal Transethmoidal Transsphenoidal
Occipital	
Convexity	Parietal Temporal

lesions, when appropriate, includes a multidisciplinary team approach, with plastic surgeons providing the expertise with craniofacial approaches and hard and soft tissue reconstruction. These lesions will be the focus of this chapter.

Congenital Masses

Encephalocele

Encephalocele refers to a neural tube defect that allows the extracranial projection of neural tissue and meninges. They may be *primary* (congenital) or secondary. *Secondary encephaloceles* are typically the result of trauma or surgery. Many theories exist, but the cause of congenital encephaloceles is not well understood. Normally, a dural projection at the fonticulus nasofrontalis contacts the surface ectoderm and later becomes encircled by the foramen cecum. This projection later involutes, leaving only a fibrous structure that fills the foramen.⁶ In sincipital and basal encephaloceles, there is a pathologic patency of the foramen cecum beyond the second month of gestation that contains cranial contents. Lesions including only meninges may be referred to as *meningoceles* and those with both meninges and neural tissue as *meningoencephaloceles*. In addition to their contents, encephaloceles may be classified according to their anatomic location (Table 39-1).

Occipital encephaloceles are the most common subtype, representing approximately 75% of all encephaloceles, followed by anterior/sincipital (12%) and basal (8%).^{7,8} The incidence of these lesions varies geographically from 1 in 5000 to 1 in 40,000 live births; the highest prevalence (particularly of the anterior and basal subtypes) is in Southeast Asia.⁹ The reason for these differences has been hypothesized to be the result of socioeconomic, nutritional, and seasonal differences.⁶

Anterior encephaloceles present as external nasal masses and basal encephaloceles as internal nasal or palatal masses that may cause nasal obstruction, difficulty feeding, or obvious cosmetic deformity.⁷ They are pale or bluish, compressible masses that transilluminate on physical examination. Encephaloceles pulsate and expand with straining, crying, or compression of the jugular vein (the Furstenberg test), which can differentiate them from other congenital midline nasal lesions. Nasendoscopy may be necessary to adequately assess the appearance and extent of these lesions. If an encephalocele is suspected, in addition to physical examination, imaging with CT or MRI can confirm diagnosis. Biopsy or aspiration should be avoided to prevent meningitis or CSF leak (Fig. 39-1).

Although these lesions are benign with no malignant degeneration potential and a slow growth rate, early surgical excision by a multidisciplinary team is the treatment of choice for encephaloceles to prevent continued facial distortion. Surgical goals include a reduction of



Fig. 39-1 Transethmoidal nasal encephalocele.

normal-appearing neural tissue into its normal anatomic location, watertight dural closure, and improved cosmesis. In patients with significant hypertelorism, orbital translocation may be necessary.^{10,11}

Glioma

Gliomas are rare lesions that are clinically similar to encephaloceles. They consist of neural tissue present at the nasal base and are thought to develop in a similar manner to encephaloceles. Gliomas, however, lack the intracranial connection of encephaloceles and exist as an isolated nest of glial tissue (Fig. 39-2). There is a male/female ratio of 3:2 with no familial predisposition.⁷ Gliomas are classified as extranasal (60%), intranasal (30%), or combined (10%).¹²

Gliomas present and should be worked up in the same manner as encephaloceles. Differences on physical examination include that gliomas are firm reddish masses that do not transilluminate. Also, findings of the Furstenberg test are negative on physical examination—there should be no change in the lesions with straining or crying. Imaging will verify the lack of intracranial connection, but 10% to 25% have a fibrous stalk with an associated bony defect.⁷ As with encephaloceles, surgical excision is the management of choice, with a goal of improving appearance and preventing infection.

Nasal Dermoid

Nasal dermoid is the most common congenital midline lesion. Although incompletely understood, these lesions form during nasal development, probably when the dural ectoderm recedes from the surface ectoderm. Nasal ectodermal elements may be pulled inward, forming a sinus tract or cyst containing ectodermal and mesodermal elements.^{13,14} Nasal dermoids are typically seen in the first several years of life; they have a male predominance and no familial predisposition.¹⁴ They account for 1% to 3% of all dermoids and 4% to 12% of head and neck dermoids.^{13,15}

Nasal dermoids present as midline nasal masses and may secrete sebaceous material or become serially infected. There may or may not be a visible punctum, but a punctum with hair growing from it is pathognomonic for a nasal dermoid. Intracranial extension has been reported in 17% to 45% of nasal dermoid lesions. Thus investigation with CT or MRI imaging, in addition to history-taking and a physical examination, is warranted.¹³⁻¹⁶ On imaging, bifidity of the

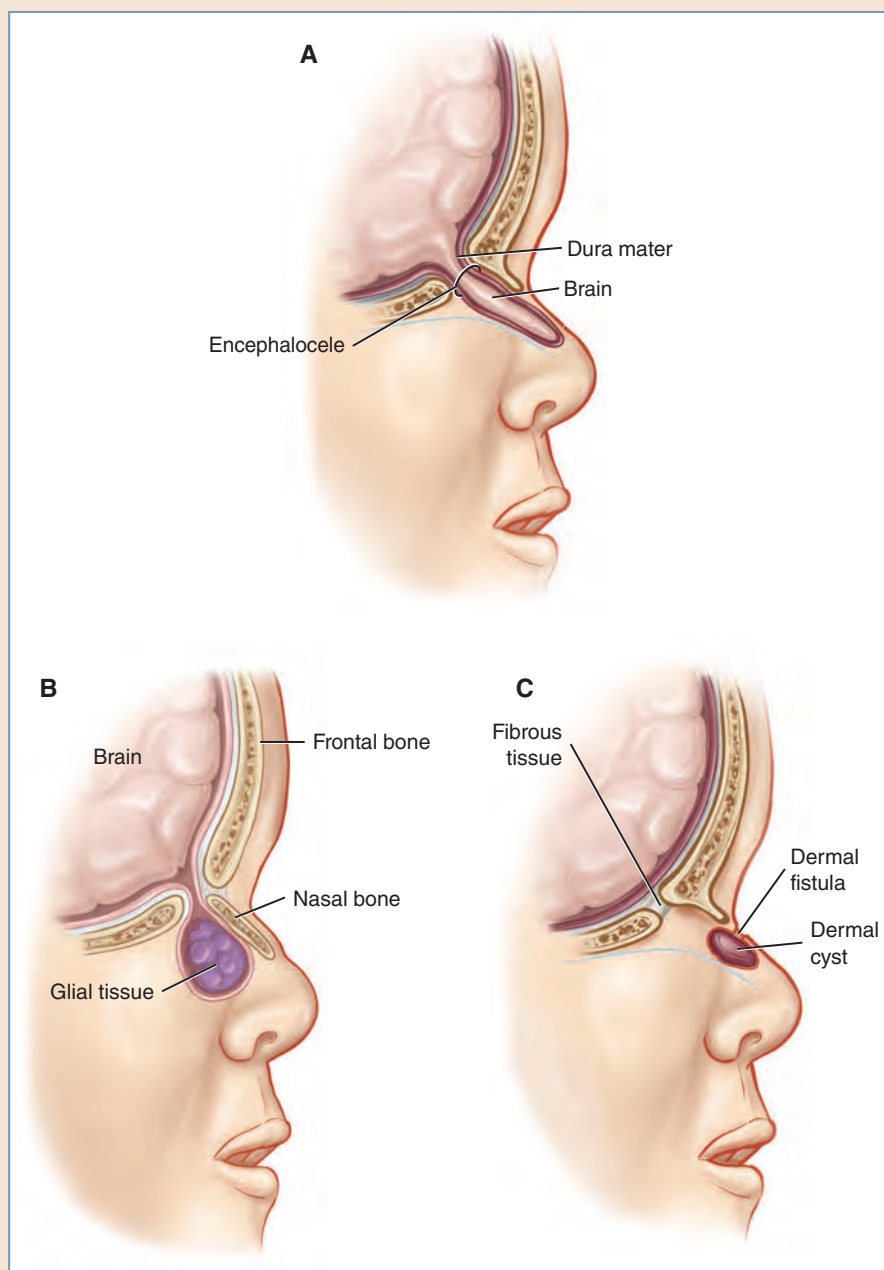


Fig. 39-2 A, Encephalocele. B, Glioma. C, Nasal dermoid.

ethmoid plate or crista galli and enlargement of the foramen cecum are highly suggestive of intracranial involvement.¹⁷

The only treatment for these lesions is surgical excision (Fig. 39-3). If imaging results lead to suspicion of an intracranial extension, some surgeons advocate intraoperative examination and clinical diagnosis of extension; others perform an intraoperative frozen-section biopsy of the dermoid stalk base to assess whether dermal or epidermal elements are present, whereas others still recommend an intracranial approach and stalk resection.^{13,14,18}



Fig. 39-3 A, This patient has a nasal dermoid with punctum. B, A probe is inserted into the tract to aid in intraoperative dissection. C, Open rhinoplasty and coronal approach. D, Intracranial connection with visible stalk after frontal osteotomy from the surgeon's perspective.

Teratomas

Teratomas are germ cell tumors derived from all three germ layers that may be mature or immature. Immature teratomas have increased potential for malignant transformation (see Chapter 37 for further discussion of teratomas).

Benign Lesions

Juvenile Nasopharyngeal Angiofibroma

Juvenile nasopharyngeal angiofibroma is a benign, locally aggressive vascular tumor. These lesions are most frequently diagnosed in adolescent males. The most commonly used staging system is based on local tumor infiltration^{13,19,20} (Table 39-2 and Figs. 39-4 and 39-5). Patients typically present with complaints of nasal obstruction, epistaxis, facial pain, facial mass, or visual disturbance. At the time of initial consultation, patients have frequently experienced symptoms for several months, and 17% to 22% have intracranial extension at the time of diagnosis.^{20,21} The lesions appear as well-circumscribed, compressible, nasopharyngeal masses that are covered with mucosa. CT, MRI, and angiography may all be used to better delineate the tumor burden.

Table 39-2 Sessions/Radkowski Juvenile Nasopharyngeal Angiofibroma Staging

Stage	Features
I	Ia: Limited to nose/nasopharyngeal vault Ib: Extension into any sinus
II	IIa: Minimal extension into pterygomaxillary fissure IIb: Full involvement of pterygomaxillary fissure $\pm$ orbital bones IIc: Intratemporal fossa $\pm$ cheek or posterior to pterygoid plates
III	IIIa: Erosion of skull base with minimal intracranial activity IIIb: Extensive intracranial erosion $\pm$ cavernous sinus

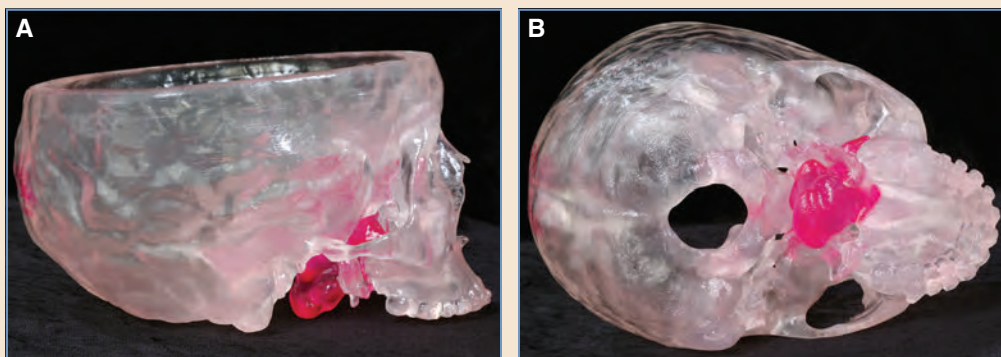


Fig. 39-4 Acrylic craniofacial model demonstrating a large juvenile angiofibroma extending into the pterygopalatine fossa (*arrow*). **A**, Lateral view. **B**, Inferior view.



Fig. 39-5 Juvenile nasopharyngeal angiofibroma expanding the pterygopalatine fossa.

Most cases do not require biopsy. However, if biopsy is performed, potential hemorrhage should be anticipated, and it should take place in the operating room.²⁰

Surgical resection is the treatment of choice for nasopharyngeal carcinoma (NPC) but may be complicated by massive hemorrhage. Even with surgical resection, there is a 18% to 22% recurrence rate.^{20,21} Preoperative embolization has been shown to significantly decrease intraoperative blood loss and is recommended especially for higher stage lesions.²²⁻²⁴ Neoadjuvant therapy with flutamide and oral antiandrogen has had variable success in shrinking tumor size.^{25,26} Radiotherapy as both adjuvant and primary treatment has shown promise in decreasing tumor size in patients with extensive disease but has not gained widespread use because of complications such as hypopituitarism, malignant transformation, and cataracts.^{27,28}

Fibrous Dysplasia

Fibrous dysplasia is a benign osseous developmental disorder in which normal bone matrix is replaced by fibrous stroma and disorganized bony trabeculae. It may be monostotic (70%), polyostotic (30%), or part of McCune-Albright syndrome.²⁹ The disease process most frequently affects the proximal femoral metaphysis and the skull base. *McCune-Albright syndrome* was traditionally described as the trio of polyostotic fibrous dysplasia, skin hyperpigmentation, and precocious puberty. More accurately, it is now known to involve fibrous dysplasia in conjunction with an extraskeletal manifestation, including several other endocrinopathies (hyperthyroidism, growth hormone excess, and Cushing syndrome).³⁰ Inheritance is sporadic and caused by an activation mutation that ultimately impairs full osteoblastic differentiation.³¹ Although these lesions are benign, there has been reported malignant transformation (less than 1%), typically to osteogenic sarcoma.³² Fibrous dysplasia is usually diagnosed in childhood, and 90% of craniofacial lesions are present before age 5 years.³³ In general, these lesions may cause deformity, pain, stiffness, and pathologic fracture. With skull base lesions, dysplastic tissue may cause cranial and optic nerve compression, with resultant cranial neuropathies and even blindness.

Workup of these lesions should include ophthalmology and audiology evaluation. CT scan is the imaging modality of choice. Findings of histologic examination in children have a typical ground-glass appearance. Fibrous dysplasia may be treated medically with bisphosphonates or surgical excision. In the case of craniofacial lesions, medical management has not proved effective. Surgical excision is suggested in the case of significant deformity or neurologic deficit.¹¹ Both conservative subtotal excision and radical resection with immediate reconstruction are acceptable surgical approaches. Adjuvant radiotherapy is not advised because of the increased risk of malignant transformation³² (Fig. 39-6).

Malignant Lesions

Nasopharyngeal Carcinoma

Nasopharyngeal carcinoma (NPC) is a malignancy of the nasopharyngeal epithelium. It is most commonly seen in adults (40 to 60 years of age), but in the pediatric population it is most commonly seen in males (10 to 20 years of age), with a male to female ratio of 1.8:1.^{34,35} The disease is rare, with an annual incidence of 0.8 to 1.3 per million in low-incidence regions and endemic regions, respectively. In North America, black children are overrepresented among pediatric cases of NPC, although it is not completely understood why. This malignancy has been classified into three subtypes by the World Health Organization³⁶ (Table 39-3). In children, type III is more prevalent, whereas type I is more common in adults.

In children NPC is treated based on the adult experience with the disease, but this malignancy varies in significant ways from NPC in adults. In children, there is a stronger association

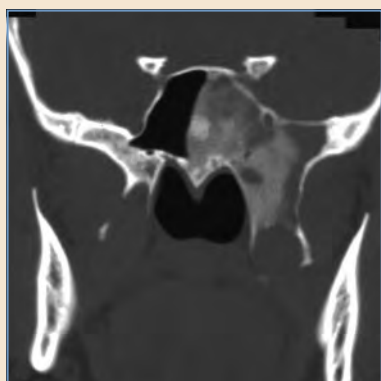


Fig. 39-6 Fibrous dysplasia lesion of the cranial base.

Table 39-3 WHO Classification of Nasopharyngeal Carcinoma

Type I	Keratinizing squamous cell carcinoma	Keratin production
Type II	Nonkeratinizing epidermoid carcinoma	Cell bundles with fusiform nuclei and scant cytoplasm
Type III	Undifferentiated carcinoma	Undifferentiated round cells with prominent nucleoli

with Epstein-Barr virus infection, a higher proportion of undifferentiated carcinoma on histologic examination, and more cases of locoregionally advanced disease at the time of diagnosis.^{34,35} NPC may present as a neck mass, nasal obstruction, epistaxis, otalgia, otitis media, headache, or neurologic symptoms as a result of skull base erosion.^{34,37} Physical examination should include assessment of cervical lymph nodes and the cranial nerve. A lesion or lymph node biopsy may confirm the diagnosis. The extent of tumor invasion may be assessed radiologically with CT and MRI. NPC is frequently unresectable because of the anatomic location and the likelihood of presentation with cervical lymph node metastases.³⁷ Treatment of NPC is based on adult protocols and consists of combination radiotherapy to the nasopharynx and affected lymph nodes and chemotherapy.³⁸

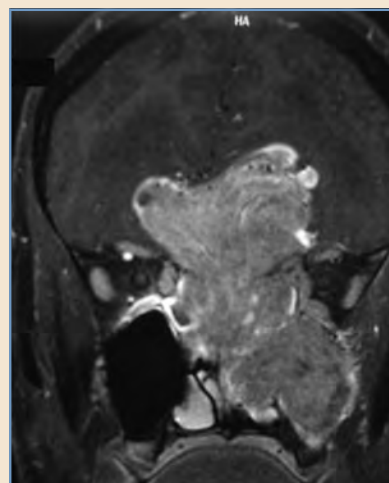
Esthesioneuroblastoma

Esthesioneuroblastoma (olfactory neuroblastoma) is a malignant, small, round cell tumor of neuroectodermal origin arising from the olfactory epithelium.³⁹ It is rare in the general population, with an incidence of 0.1 per 100,000 in children younger than 15 years of age.⁴⁰ Despite that, it is the most common cancer of the nasal cavity in the pediatric population. Esthesioneuroblastoma has a bimodal age distribution, with peaks in children ages 11 to 20 and adults ages 51 to 60.⁴¹ There does not appear to be a sex predisposition. Staging of these lesions was originally based on the anatomic extent of the disease, and this is still the most commonly used staging system, although more recent TNM staging has also been adopted^{42,43} (Table 39-4). The most common complaints of patients with esthesioneuroblastoma include nasal obstruction and epistaxis.

Table 39-4 Kadish-Morita Staging of Esthesioneuroblastoma

Stage	Feature
A	Tumor limited to nasal cavity
B	Tumor limited to nasal cavity and paranasal sinuses
C	Tumor extends beyond nasal cavity and paranasal sinuses
D	Tumor with neck or distant metastases

Fig. 39-7 An MRI of esthesioneuroblastoma invading the anterior skull base, frontal lobe, nasal cavity, and orbit.



On examination, these lesions appear to be reddish or gray tumors arising in the upper nasal fossa and may bleed easily with manipulation (Fig. 39-7). Workup of these lesions includes imaging with CT, MRI, and biopsy. Single, double, and triple modality (surgery, radiation, and chemotherapy) treatment protocols have been used, and surgical excision plus radiotherapy has the highest cure rate.³⁹ Regardless of therapy, prognostic factors include histologic grading and the presence of cervical lymph node metastases.³⁹

Rhabdomyosarcoma

Rhabdomyosarcoma is the most common soft tissue sarcoma in children. Thirty-five percent of those patients who have a diagnosis of rhabdomyosarcoma present with a head and neck tumor (Fig. 39-8) (see Chapter 37 for further discussion of rhabdomyosarcoma).

SURGICAL APPROACHES

Advances in surgical approaches to the anterior cranial base have dramatically improved our ability to access these lesions and improve patient prognosis. Various techniques may be used to expose lesions of the cranial base. In general, both intracranial and extracranial approaches may be used. An exclusive intracranial or extracranial approach may be sufficient in cases of small lesions in a favorable location. Most of the time, however, a combined intracranial and extracranial

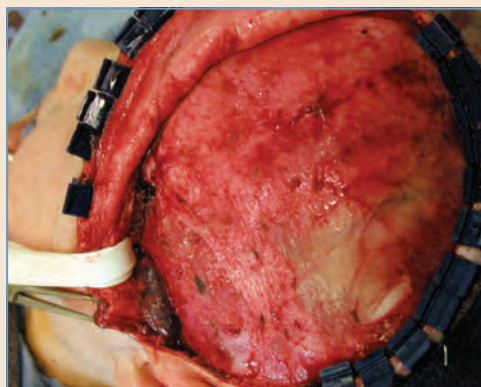


Fig. 39-8 Rhabdomyosarcoma of the infratemporal fossa.

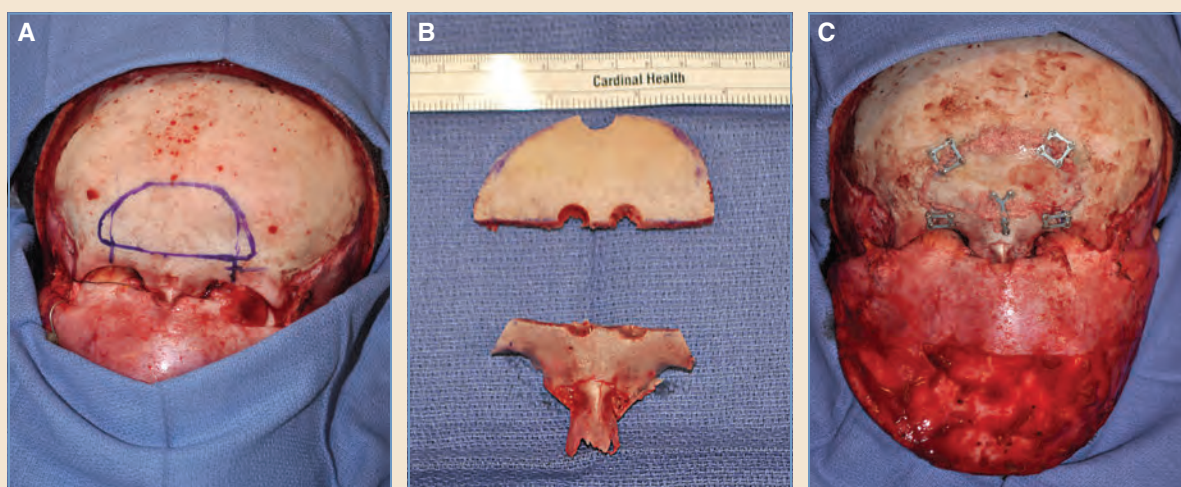


Fig. 39-9 A, Markings for nasofrontal osteotomy. B, Bone flaps. C, Bone flaps replaced with bone graft.

approach is necessary. Approaches may be viewed in terms of soft tissue handling and osteotomies used to access the cranial base.

Soft tissue exposure may be achieved with a combination of coronal, transfacial, and gingivobuccal sulcus incisions. A standard coronal incision provides access to the frontal bone, orbital roofs, and nasal bone. It is most commonly used to access the frontal bone for intracranial exposure through a frontal osteotomy or superior orbital osteotomy. A pericranial flap is frequently raised to obliterate dead space or to reestablish anatomic boundaries after resection (Fig. 39-9). A transfacial open rhinoplasty or lateral rhinotomy/Weber-Ferguson approach provides wide exposure of the maxilla, orbital floor, and zygoma. With this soft tissue exposure, a maxillectomy, palatal split, and LeFort I osteotomy may be performed to approach the cranial base. A LeFort I should be performed with caution in children with mixed dentition, because it may damage unerupted tooth buds. Transoral access with an upper gingivobuccal sulcus incision allows a facial degloving technique that avoids incisions on the face and provides access to the maxilla, orbital floor, and zygoma. After degloving, maxillectomy, ethmoidectomy, and LeFort I osteotomy may be performed. Finally, the endoscopic endonasal approach to the anterior skull base has become increasingly performed as technique, imaging, and instrumentation have improved. It may be

used alone, particularly for extradural lesions of the skull base. It may also be used in conjunction with craniotomy.

CONCLUSION

Cranial base masses are best managed by a multidisciplinary team, particularly malignant lesions that require a combination of medical and surgical management and long-term follow-up. Cranial base masses have multiple causes, including congenital, benign, and malignant lesions. Thorough investigation, including the use of CT and MRI scanning, should be undertaken to make a diagnosis.

Encephaloceles, nasal dermoids, and gliomas are congenital lesions that may be diagnosed at birth. As with most congenital lesions, some remain undiagnosed until they become secondarily infected in childhood or later. Despite the name, benign tumors can be extremely destructive. Depending on their location, they can cause distortions of the overlying craniofacial skeleton and soft tissue or exert mass effect, causing visual or olfactory disturbance or cranial nerve neuropathy.

Malignant lesions may have the same effects, with the added risk of metastasis. When surgical intervention is indicated for these lesions, the most common approach remains the combined intracranial and extracranial approach, typically performed by a combination of plastic surgeons and neurosurgeons, which again underscores the importance of a team approach in the care of these patients.

KEY POINTS

- A multidisciplinary surgical team is the benchmark for treating patients with anterior cranial base lesions.
- Encephaloceles and gliomas are characterized by extracranial projection of neural tissue and are managed with surgical excision.
- Unlike dermoid cysts elsewhere, nasal dermoid cysts may have intracranial extension and should have imaging performed as a part of their workup.
- Benign cranial base tumors may be extremely locally destructive.
- Malignant cranial base tumors are rare in children. Management may be based on adult protocols despite differences in adult and pediatric disease characteristics.
- Typically, intracranial and extracranial approaches are combined to access anterior cranial base lesions.
- An endoscopic endonasal approach to the anterior cranial base has become increasingly popular.

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Pediatric Craniofacial Fractures

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The American Academy of Pediatrics defines pediatric patients as those between birth and 21 years of age. The wide variation in function, anatomy, and growth potential that occurs within this age range makes any single approach to facial fractures in pediatric patients challenging. Even though fractures of the craniofacial skeleton are uncommon in children compared with adults, they are nonetheless frequently more difficult to treat because of significant anatomic differences and the potential for long-term growth and development.¹⁻⁶ Although the goals of treatment are the same as those for adults, the approaches to pediatric patients with facial trauma are often dramatically different.⁷⁻⁹ Even after years of advancement in craniofacial trauma treatment, many questions regarding pediatric patients remain unanswered. In this chapter, we will review the up-to-date literature, delineate the differences between adult and pediatric fracture patterns and management, and provide guidelines for the reconstruction of pediatric craniofacial fractures.

GROWTH AND DEVELOPMENT OF THE CRANIOFACIAL SKELETON

Understanding the centers of growth and development of the craniofacial skeleton is crucial to the ability to treat pediatric craniofacial trauma. Within the craniofacial skeleton, there are significant regional differences in growth patterns that continue to change throughout development. At birth, the cranial/facial proportion is 8:1. This ratio becomes 4:1 at around 5 years of age and 2:1 in adulthood.¹⁰ The growth of the neurocranium is 25% complete at birth, 75% by 2 years, and 95% by 10 years of age. In comparison, facial growth is only 65% complete at 10 years of age.¹⁰⁻¹³ Cranial growth is a continuous process that occurs in a defined period of time

and is essentially dictated by brain expansion under the influence of the cranial sutures. Indeed, the brain's volume triples by the end of the first year of life. The sutures gradually narrow, and the fontanelles are closed in sequence. This rapid growth occurs until 3 years of age and then dissipates into adulthood.¹⁰

In contrast, facial growth demonstrates a drastically different pattern of discontinuous growth until puberty is complete. Multiple mechanisms, such as facial synchondroses, sutures, and apposition and resorption, play roles in this process. At 3 months of age, the facial dimensions are 40% of those in adults, and they become 70% by 4 years and 80% by 5 years of age.¹⁰ At approximately 5 years, growth slows dramatically until the beginning of puberty, at which point accelerated growth occurs mediated by increased hormonal input. At 17 years, facial growth slows down again and eventually halts.

Upper facial skeletal growth occurs as a result of cerebral and ocular growth. Orbital expansion is complete by 6 to 8 years of age¹⁰; the frontal sinus becomes visible by 5 years and obtains complete aeration well after puberty. Midfacial growth is associated with dental development. By 2 years of age, transverse maxillary growth is near complete, whereas palatal and midline maxillary suture growth ceases between 8 and 12 years. At 12 years of age, pneumatization of the maxillary sinus occurs, along with dental eruption.^{10,12,13} Lower facial growth also shows sequential growth. The mandibular symphysis fuses completely by 2 years of age when the deciduous teeth are erupting. The primary growth centers of the mandible are thought to be the condyles, and they contribute to vertical growth, activated by muscular activities. These activities place stress on the periosteum, which in turn causes apposition and resorption of bone, creating mandibular growth in all other directions.^{9-11,13,14}

EPIDEMIOLOGY

Although the incidence of facial trauma is higher in children than in adults, the incidence of pediatric craniofacial fractures is much lower.^{15,16} Rowe² reported that only 1% of all facial fractures occurred in children less than 6 years of age. His findings have been echoed by McCoy et al,¹ Panagopoulos,¹⁷ Kaban et al,¹⁸ and others. However, most of these reports were made before CT was used to evaluate pediatric facial trauma, and therefore they likely underestimated the incidence of pediatric facial fractures.¹⁹ Several factors contribute to the overall low incidence of pediatric craniofacial fractures. First, infants and young children usually live in a protected environment; therefore the chance of trauma with significant impact is lower.¹⁰ As children grow older and begin to play independently and participate in sports, the risk of facial trauma increases. Second, unlike adults, children have a lower facial mass/cranium ratio and are therefore much more likely to sustain skull fractures and head injuries than facial fractures.^{10,20,21} Third, the pediatric facial skeleton is quite elastic because of immaturity, lack of sinuses, a higher percentage of cancellous bone, and the presence of cartilaginous sutures and growth centers. These factors allow pediatric facial bones to absorb more energy during impact without fracturing than adult facial bones.

The pediatric population varies in its distribution of facial fractures. For example, boys are twice as likely as girls to sustain facial fractures. In addition, the 6- to 12-year-old age group is more prone to facial fractures than any other age group. This is likely because the presence of mixed dentition and a higher tooth-to-bone ratio weakens bone.²² Other variations in fracture distribution with respect to age are likely associated with the decrease of the cranial/facial ratio that results in a more prominent face as the child grows.^{11,16,18} In addition, as the facial skeleton matures it becomes increasingly mineralized and pneumatized and therefore more prone to fracture. As pediatric patients age, there is a downward shift (superior-inferior) of the craniofacial trauma fracture pattern. Frontal skull and orbital fractures have a higher incidence in the new-

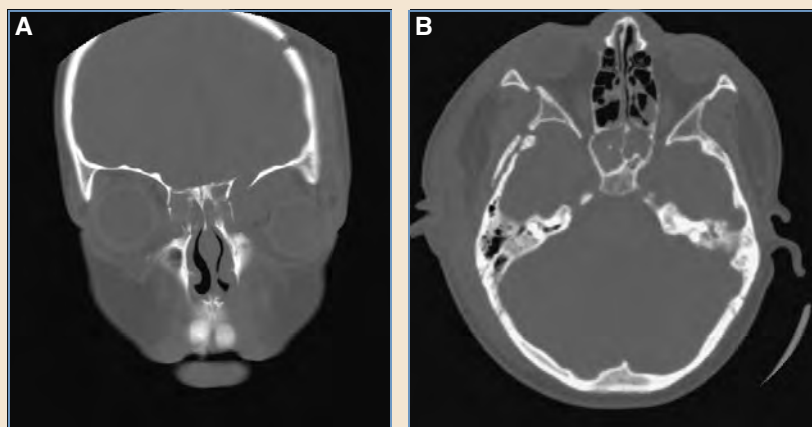


Fig. 40-1 A, Coronal CT scan showing a left-sided oblique craniofacial fracture pattern. B, Axial CT scan showing a right-sided oblique craniofacial fracture pattern.

born to 5-year-old age group, whereas midface and mandibular fractures have a higher incidence in the 6- to 16-year-old age group.²³ The nose and mandible account for most reported facial fractures in older children, which is likely because of their prominence.¹⁹ McCoy et al¹ and Bales et al⁷ found that mandible fractures were more common than nasal fractures, whereas Kaban et al¹⁸ reported the opposite. Midface fractures, including orbital, zygomatic/malar, and maxillary, are less common, as reported by both Kaban et al¹⁸ and McCoy et al.¹ In a recent analysis of 772 fractures in patients presenting to the emergency room of a level 3 trauma center, Grunwaldt et al²³ found that the highest fracture rate was in older patients, with 48% of fractures occurring in 12- to 18-year-old patients. In addition, younger patients were more likely to have frontal and orbital fractures caused by falls or motor vehicle collisions (MVCs), whereas older patients were more likely to sustain maxillary, nasal, and mandible fractures as a result of violence/assault, MVCs, and sports.²³⁻²⁵

When subjected to equal force, children develop incomplete greenstick fractures more often than adults.^{19,26,27} This is because the increased elasticity of the less mineralized and pneumatized pediatric skeleton and greater cancellous-to-cortical bone ratio add much more flexibility to the mandible and maxilla. These factors also result in unique fracture patterns seen in children; for example, it has been reported that more oblique craniofacial fracture patterns are found in immature facial trauma patients²⁷ (Fig. 40-1). The classic LeFort fracture patterns that are seen in mature craniofacial skeletons are rarely found in immature patients.

Causative mechanisms of pediatric facial fractures include motor vehicle accidents (MVAs; 50%), falls (23%), and sports (15%).⁶ Assaults represent a much less contributing factor⁴; however, violence-related facial fractures are more likely in older male patients of lower socioeconomic status.²⁴ There is a marked seasonal variation of pediatric facial fractures, which peaks in July. Recently, all-terrain vehicles (ATVs) have become an increasingly more common cause of pediatric facial fractures.²² Together, MVAs and ATV accidents cause more severe midface fractures than any other mechanism. This is especially evident in adolescents.²² Fortunately, child abuse is a rare cause of pediatric facial fractures, but it should be seriously considered as a cause in patients with associated unexplained long bone fractures.

DIAGNOSIS

Diagnosis of pediatric facial fractures is usually challenging. One must maintain a high degree of suspicion, especially in the presence of injuries to major organ systems. It is not unusual to have a patient who is unable to cooperate accompanied by anxious parents in the emergency department, which may necessitate sedated examination when safely possible. A history must be carefully obtained from both the parents and any witnesses to the incident, along with emergency medical support personnel. Preinjury photographs of the child, dental records, and models from the child's dentist and orthodontist are often helpful when treating children with facial fractures.

Plastic surgeons must work closely with the physician team to coordinate the care of a child with multiple injuries, following the Advanced Trauma Life Support protocol. The examination of the cervical spine is especially important in children with trauma. There is a reported 10% associated risk of concomitant craniofacial trauma and cervical spine injury.²⁸

Examination of a patient with craniofacial trauma should begin by observing the overall condition of the patient. Alertness and orientation should be carefully documented. A low threshold should be maintained for neurosurgical consultation in children with craniofacial trauma, because 75% of frontal sinus fractures are accompanied by loss of consciousness,¹⁹ and close to half of patients with facial fractures have accompanying neurologic injuries.²³ The airway should be thoroughly evaluated. Endotracheal intubation in these patients is uniquely challenging, because cervical spine clearance is often not entirely possible and may also be complicated by fractures of the mandible or midface. Pediatric anesthesia support for nasal intubation over a flexible endoscope may be performed with limited cervical spine motion and may be the best way to establish a nonsurgical airway.²⁸ If all else fails, tracheostomy or cricothyrotomy can be used.

Physical examination begins with the head. Careful palpation of the skull should be carried out after adequate cleansing of any scalp wounds, and this may require local anesthesia. Any scalp lacerations must be washed out and carefully examined for underlying skull fracture. Next, attention is given to the superior orbital ridge. Fractures in this region can often cause paresthesia of the forehead and may be associated with traumatic ptosis of the brow. It is important to realize that in young patients without a frontal sinus, forehead and supraorbital ridge fractures are essentially anterior cranial base fractures.

Orbital examination is extremely important, because there is a significantly higher percentage of ocular trauma and blinding injuries in this population compared with adults. Studies have reported as high as a 3% blindness rate in children with orbital fractures.²⁹ Consultation with a pediatric ophthalmologist should always be obtained when children present with periorbital trauma. Subconjunctival hematomas are pathognomonic for orbital fractures, because the conjunctiva is contiguous with the orbital periosteum; therefore patients with these conditions must be assumed to have an orbital fracture until a CT scan is obtained. Bilateral vision should be carefully documented, and when possible, evidence of afferent papillary defects is verified and documented. Extraocular movements should be tested voluntarily in conscious patients, and forced duction testing should be performed with unconscious patients and all patients undergoing surgery. Total ophthalmoplegia, along with upper eyelid ptosis and paresthesia in the V1 distribution, indicates the presence of superior orbital fissure syndrome. If blindness is present in addition to these findings, then orbital apex syndrome is diagnosed. Both these conditions are ophthalmologic emergencies that may require urgent surgical decompression and high-dose steroid therapy. The medial canthal tendons should be tested in patients with periorbital trauma. This is done with the bow-string test, which is performed by pinching the eyelids and distracting them laterally to check the stability or mobility of the medial canthus. Intercanthal distance

should be measured and documented. Any periorbital lacerations medial to the lacrimal puncta indicate potential damage to the lacrimal drainage system.

Examination of the nose begins with the nasal root and continues caudally. Deviation of nasal bones should be evaluated, as well as compressibility of the nasal dorsum. Any rhinorrhea or nasal discharge should raise suspicion of a CSF leak, therefore prompting a CT scan and consultation with the neurosurgical staff. A thorough intranasal examination should be carried out with a nasal speculum and an adequate light source, and any septal hematomas should be treated promptly.

Examination of the midface begins with the inferior orbital rims. Fractures in this region are often associated with cheek paresthesia and a step deformity that can be felt with manual palpation. The entire zygoma should be palpated. Fractures of the zygoma often result in lateral canthal dystopia and a loss of malar prominence. Midface stability should be tested by attempting manual distraction of the maxilla while stabilizing the head; any independent movement of the maxilla may indicate a midface fracture. However, if the patient has an obviously mobile midface, one should refrain from manipulating it more, because this may cause optic nerve trauma. Occlusion is also an important diagnostic maneuver when one is evaluating the midface, and abnormal occlusal findings should prompt further radiologic studies. During examination of the midface, attention should also be given to the ears. Hemotympanum, as well as a mastoid hematoma (Battle's sign), may indicate a basilar skull fracture. External trauma, including subperichondrial hematomas, requires prompt treatment.

Lower craniofacial skeleton examination begins with the mandible; the patient's own subjective assessment of occlusion is obtained whenever possible. Subjective malocclusion is a sensitive indicator of occlusal disorders. Objectively, malalignment of the dental arch and open bites are indicative of jaw fractures (Fig. 40-2); however, patients with mixed dentition often have such preexisting malocclusions, which is normal for their stage of development. Angle's classification should be used to document findings, and any evidence of malocclusion should prompt obtaining a panoramic film and/or a CT scan. Chin paresthesia is found often in patients with parasymphysal fractures. Palpation of the mandible starts at the temporomandibular joint (TMJ) and ends at the symphysis. A high index of suspicion must always be maintained with children, because pain during palpation is often the only indicator of facial fractures.

Diagnostic imaging to evaluate pediatric facial fractures has evolved, and in modern practice sophisticated CT algorithms have limited the role of plain radiography, because the pediatric facial skeleton is immature and has a high cancellous/cortical bone ratio. Fine-cut spiral CT scans



Fig. 40-2 Malalignment of the mandibular dental arch, which is indicative of fracture.

from skull to mandible should be used whenever possible. True coronal images are ideal, although if unable to clear the cervical spine, reconstructed coronal images, based on the axial CT scan, can often suffice. Three-dimensional reconstructions of fine-cut CT scans are exceedingly useful for diagnosing fractures and treatment planning. The Panorex is the single plain film that remains useful to evaluate pediatric patients who are able to cooperate for the study.

TREATMENT OF FACIAL FRACTURES: UNIQUE FACTORS

Managing pediatric facial fractures is often difficult, because the goal of obtaining the best possible reduction and fixation must be balanced with potential growth disturbances associated with open surgical approaches to the pediatric facial skeleton.¹⁰ Evidence of facial growth restriction after wide subperiosteal dissection has been reported³⁰; however, Bartlett et al³¹ reported no growth disturbances after wide undermining of the frontoorbital region, such as from the treatment of craniosynostosis.

Different areas within the facial skeleton demonstrate different healing properties. In a growing child, the condyles of the mandible have an excellent ability to remodel, and therefore a conservative approach may often be taken with pediatric condylar fractures. It is prudent to accept minor occlusal disturbances in a growing child while the surgeon attempts to treat fractures conservatively, because these may be corrected with future orthodontic treatment. Other areas, such as the orbit, may not tolerate malunion so well and therefore may require adequate exposure for internal fixation. It is imperative that surgeons clearly communicate to parents the potential risks of growth disturbances that the fracture itself, along with its surgical correction, may impose. In addition, there is an increased risk to unerupted and developing tooth buds in the jaws of pediatric patients. Communication helps parents to understand the need for long-term follow-up.

Another unique controversy in the treatment of pediatric facial fractures is the use of alloplastic materials and rigid fixation. The advantages of titanium miniplate and screw fixation include increased control and decreased mobility of bony fragments and improved contour postoperatively.³² The disadvantages of using these plates and screws include hardware migration and potential growth restriction.¹⁵ Yaremchuk et al³³ studied the effects of plate and screw fixation on the growing craniofacial primate skeleton with wire fixation, limited miniplate and screw fixation, and extensive miniplate and screw fixation. They found that a small but measurable visible change occurred in the growing primate skull after miniplate and screw fixation. The magnitude of these changes appears to increase with an increased amount of fixation hardware used.³³ Many have also noted that metallic hardware in a growing skull can translocate to the endocranial positions after craniofacial surgery (Fig. 40-3). Metallic wires and hardware have become embedded in the dura; however, to date this has only posed potential hazards. Because of these findings, many surgeons feel that only absorbable hardware is indicated for pediatric patients.³⁴ In the past 15 years, resorbable fixation systems have become widely available and allow reasonable fracture fixation in the short term without limiting growth in the long term.³⁴

Pediatric patients with facial fractures often do not display the standard LeFort fracture patterns found in adult patients.³⁵ This oblique fracture pattern is often seen with high-velocity trauma, as described by Moore et al²⁷ (see Fig. 40-1). This fracture pattern runs obliquely from the cranial base to the frontal bone with extension into the orbit and maxilla. Extension to the mandible is rare. This pattern is supposedly caused by the absence of rigid bony buttresses that are present in adults.¹⁰

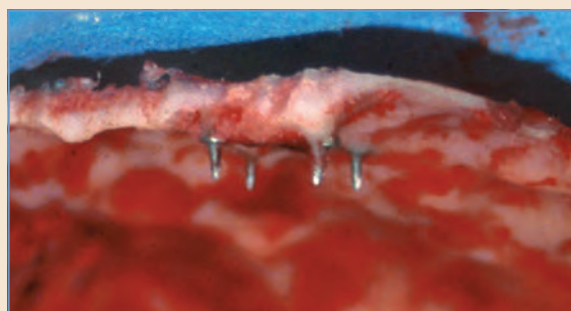


Fig. 40-3 Translocation of metallic plate and screws to the endocranial side of the skull.

TREATMENT OF FRACTURES

Skull and Forehead

Children have larger cranium-to-face ratios than do adults. This is in addition to the more elastic nature of pediatric bone and the lack of a developed frontal sinus,⁵ which begins its pneumatization around 5 years of age and completes the process at about 20 years of age.³⁶ Because of this, children who sustain significant force to the skull frequently have fractures extending to the cranial base.⁸ Mann et al³⁷ reported that in 1297 pediatric skull fractures, the incidence of associated intracranial injuries was directly related to age. Whereas children younger than the age of 2 years with open skull fractures had a negligible chance of developing intracranial hematomas, 15-year-old children had an incidence of intracranial hematoma equivalent to adults.³⁷ Bonfield et al³⁸ revived 897 pediatric skull fracture patients, the vast majority of which did not require intervention. Factors associated with the need for intervention included direct blow to the head with an object or an MVC. Although children with head injuries generally have a better chance of survival than adults, the long-term sequelae are more significant.

Skull fractures often occur in an oblique orientation in pediatric patients, with a fracture originating at the site of impact, extending to the supraorbital foramen, and progressing to involve the orbit and zygoma, resulting in an oblique craniofacial fracture.¹⁹ Oblique fracture patterns can be exceedingly difficult to treat, because a craniotomy is required to reduce the often depressed and impacted frontotemporal skull fracture that is contiguous with the facial fracture (see Fig. 40-1). The most common mechanisms of pediatric skull fractures are MVAs, followed by falls and fights. There is a 20% correlation between long bone injuries and skull fracture in this population.

Diagnosis of skull, forehead, and cranial base fractures should be done by a careful physical examination, followed by a CT scan. A CT scan is necessary because of the high incidence of associated intracranial injuries. Any scalp laceration should be explored under sterile conditions to determine bony continuity and to rule out skull fracture. Eyelid hematoma, acute hearing loss, hemotympanum, or cranial nerve palsy are often present with basilar skull fractures.

Unique to pediatric craniofacial fractures is the *growing skull fracture*.³⁹⁻⁴¹ A growing skull fracture is thought to occur when the fracture is associated with a dural tear. The dural diastasis allows brain pulsations to slowly separate the fracture edges, resulting in nonunion and “growth” of the fracture. One site for growing skull fractures is the frontal bone with extension into the supraorbital ridge, resulting in orbital changes such as bony orbital expansion, vertical orbital

dystopia, and pulsatile exophthalmos.⁴¹ Contrary to previous reports,⁴² a recent series published by Losee et al⁴³ demonstrated growing skull fractures in more than 4% of all pediatric orbital fractures and 15% of orbital roof fractures. For this reason, long-term follow-up is critical in this patient population (Fig. 40-4).

Frontal bone, frontal sinus, and cranial base fracture treatment should achieve four goals^{19,44}:

1. Isolation of intracranial structures (for example, separation of the brain from the nose)
2. Cessation of CSF leakage
3. Prevention of posttraumatic infections (for example, pyocele and mucocele)
4. Restoration of facial contour

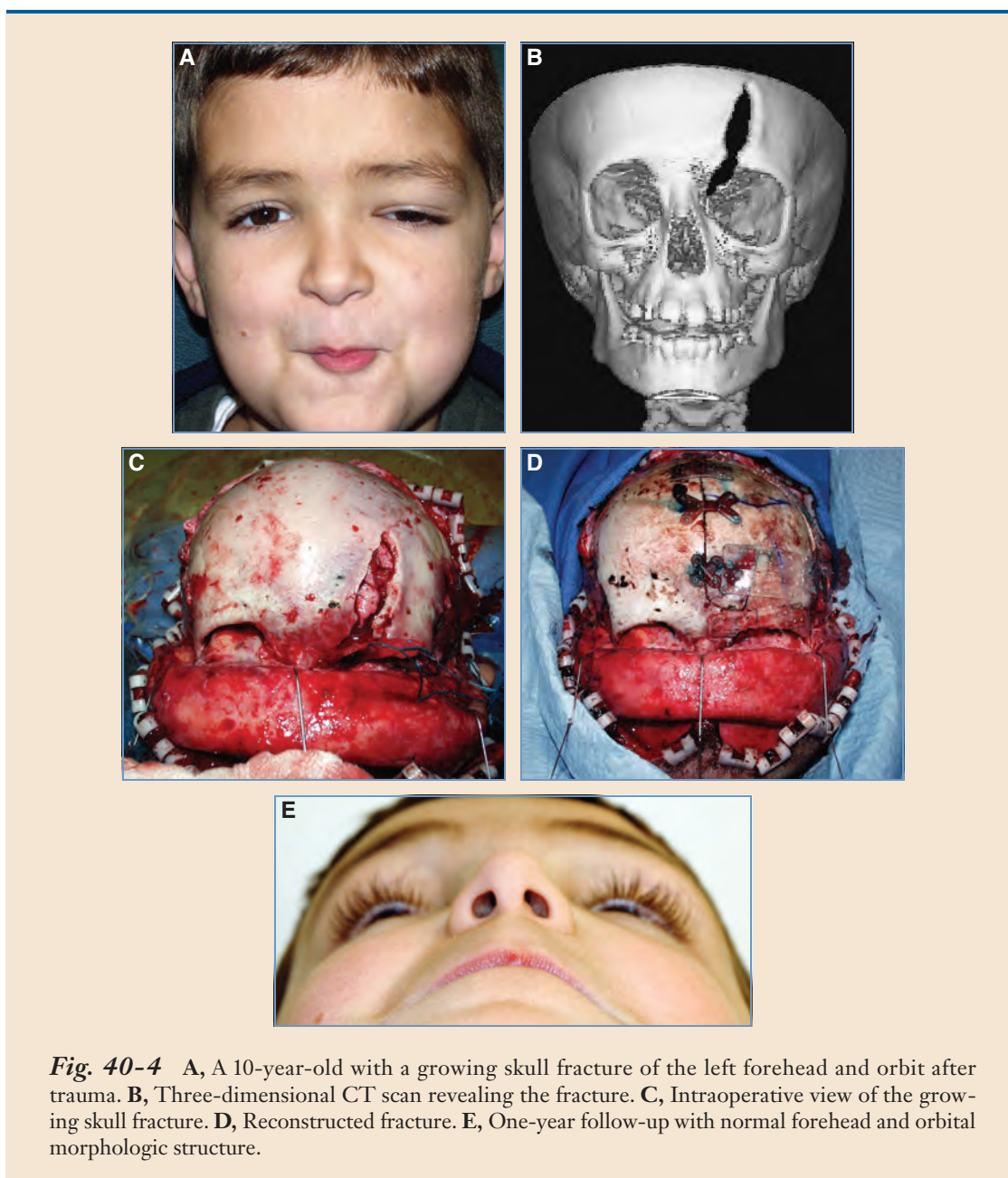


Fig. 40-4 A, A 10-year-old with a growing skull fracture of the left forehead and orbit after trauma. B, Three-dimensional CT scan revealing the fracture. C, Intraoperative view of the growing skull fracture. D, Reconstructed fracture. E, One-year follow-up with normal forehead and orbital morphologic structure.

Indications for surgical intervention include persistent CSF leakage (more than 4 days), intracranial/extracranial hematoma, significant bony displacement, and deformed facial contour. Treatment of these fractures often requires the assistance of neurosurgical colleagues in a combined approach. Nondisplaced or minimally displaced frontal bone fractures are often treated nonoperatively. Displaced fractures are treated with a coronal incision approach, access by craniotomy, and fixation with either resorbable plating systems in young patients or wires or titanium plating systems in those patients near adulthood (Fig. 40-5). When the frontal sinus is pneumatized, the treatment should be specific to address each fracture pattern and to ensure either a “safe sinus” by confirming adequate drainage and prevention of future mucoceles or obliteration/cranialization of the sinus. With advances in the treatment of frontal sinus disease, placement of frontonasal duct stents may potentially help to maintain patency. For fractures involving the frontonasal ducts, the frontal sinus is usually obliterated or cranialized. All sinus mucosa is carefully removed with loupe magnification, methylene blue dye, and a burr.¹⁹

Incomplete mucosal removal increases the risk that mucocoele, mucopyocele, and brain abscesses will occur several years after the initial surgery. When the sinus mucosa is completely removed, the space is left either empty or obliterated. Various autologous materials have been used to obliterate the frontal sinus, including bone, fascia, pericranium, and fat. We feel that the

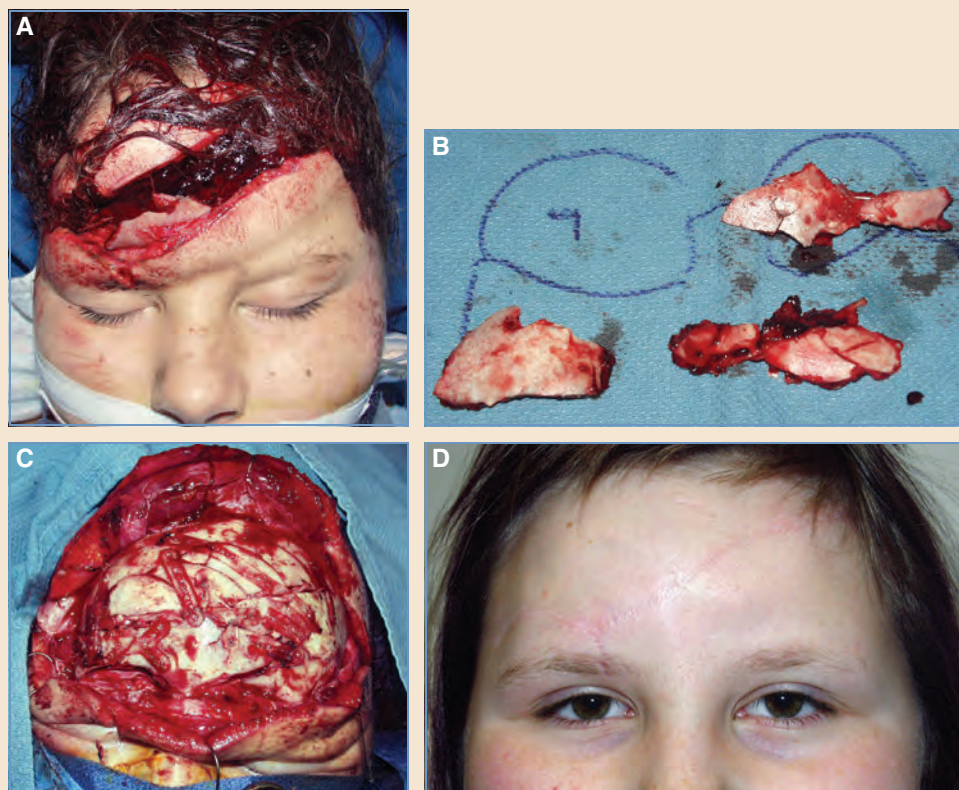


Fig. 40-5 A, A 13-year-old with a frontal bone fracture after an ATV accident. B, Frontal bone pieces mapped out. C, Reconstruction with absorbable plates. D, Six-month follow-up after reconstruction.

procedure of choice for sinus obliteration involves the use of cancellous bone from the iliac crest, taking care to obliterate the frontonasal ducts. When there are comminuted fractures of the posterior wall of the sinus, with or without CSF leaks, the sinus is often cranialized. Cranialization of the sinus includes removal of the posterior wall, complete removal of all mucosa, blocking the frontonasal ducts with bone grafts, and completing the isolation of the intracranial contents with a pericranial and/or a galeal flap.

If the nasocranial defect is large, an additional inferior or nasal approach can be used to separate the nose from the brain. Galeal grafts or AlloDerm (LifeCell Corp.) can be endoscopically placed in the superiormost nasal vault recess at the defect with tissue adhesives and glues. A course of nasal packing completes this technique. The incidence of meningitis after surgical treatment of frontal sinus fractures in pediatric patients is about 5%.⁴⁵ Long-term follow-up of children with frontal bone/sinus fractures is required to ensure that their frontal sinuses have adequate drainage as they grow. Imaging is often suggested for follow-up visits. Although routines may vary, it is reasonable to repeat a CT or MRI scan every 3 to 5 years to rule out mucocele formation.

For pediatric patients with a cranial injury that involves bone loss, the task of reconstruction is difficult. The procedure of choice for calvarial reconstruction remains the use of split calvarial grafts; however, this option is unavailable to younger patients. The diploic space between the inner and outer tables of the calvarium begins to be evident at 3 years of age and can be fully apparent by 9 years of age. Therefore, when harvesting split calvarial bone grafts in children younger than 9 years of age, a full-thickness cranial bone graft should be obtained, and the split should be performed on the back table if possible.

After the age of 9 years, *in situ* harvesting becomes much safer. The ribs and ilium also provide a source of autologous bone for calvarial reconstruction. Usually multiple ribs are harvested and split. Each segment is then contoured with bone-bending forceps and applied to the defect. However, rib grafts are plagued by high rates of resorption.⁴⁶ Although synthetic materials abound (for example, methylmethacrylate and titanium), it is best to use autologous material when possible in a growing child.⁴⁷ When synthetic materials are used, the age and growth potential of the child must be considered. This is only significant for calvarial reconstruction in the 2- to 10-year-old age group when children have lost their osteogenic potential and their diploic space is not developed enough for splitting.

In our experience, a bilaminate construct composed of absorbable mesh placed both intracranially and extracranially, interposed with a compact layer of demineralized bone matrix and chips or shavings of bone graft and used in primary craniofacial surgery, has led to bony regeneration, providing a stable reconstruction in patients of this age group. The surgeon must recognize that this technique is successful for primary calvarial defects with nonscarred or injured dura or scalp. In the compromised and/or scarred environment, this technique has demonstrated limited success⁴⁸ (Fig. 40-6). In addition, we have increasingly used custom porous polyethylene implants to reconstruct large cranial defects when soft tissue quality was uncompromised⁴⁹ (Fig. 40-7).

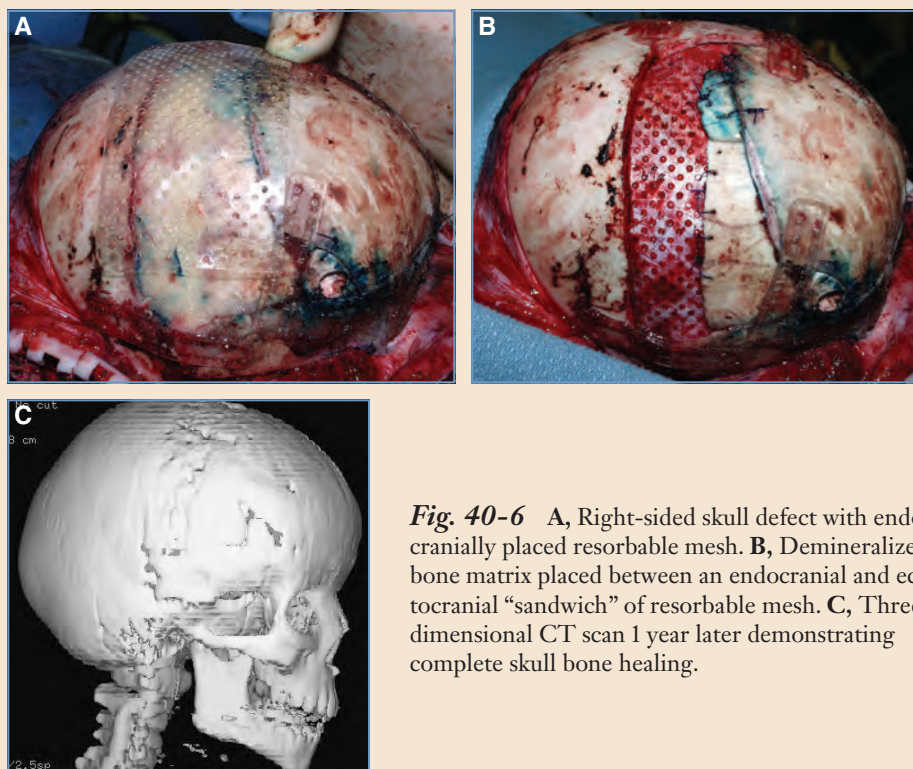
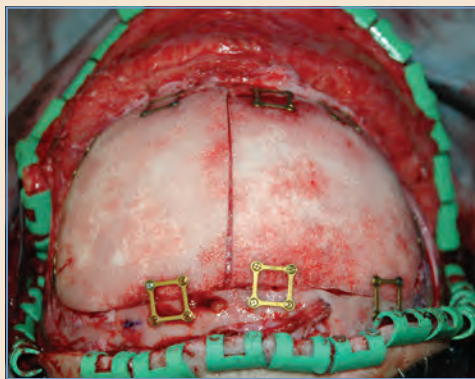


Fig. 40-6 A, Right-sided skull defect with endocranially placed resorbable mesh. B, Demineralized bone matrix placed between an endocranial and ectocranial “sandwich” of resorbable mesh. C, Three-dimensional CT scan 1 year later demonstrating complete skull bone healing.

Fig. 40-7 Medpor implant.



Orbital Fractures

Pediatric orbital trauma is rarely isolated and is often associated with zygomatic fractures.¹⁹ The orbit reaches adult size near the age of 7 years, and both the small maxillary sinuses and flexible bone contribute to a lower incidence of blowout fractures in children. Tooth buds provide support to the orbit in very young children, particularly in the inferior medial region of the floor. The most common causes of pediatric orbital fractures are MVAs; child abuse is second in prevalence. The separation of the frontozygomatic suture in the lateral orbital wall is quite common

in children, whereas it is uncommon in adults.¹⁹ In addition, the frontal sinus is absent before the age of 7 years. Therefore traumatic energy is transmitted to the floor of the anterior cranial vault, which is the orbital roof, causing isolated orbital roof fractures. When there is an isolated orbital floor fracture, children are prone to trapdoor-type fractures, in which the inferior rectus or inferior oblique muscle becomes trapped beneath the fractured segment, which has “snapped back” into position. This is one of the true surgical emergencies in pediatric facial trauma management (Fig. 40-8). Immediate surgical exploration and release of the trapped muscle are critical, because the muscle has an increased risk of ischemic injury in a relatively short period of time and can suffer permanent damage if not treated in a timely manner. Although pediatric orbital fractures are less common than in adults, there is a significantly higher incidence of blinding injuries in the pediatric population.²⁹

Therefore a higher index of suspicion for orbital injuries should be maintained for children with any evidence of periorbital trauma, and such patients should undergo a thorough ophthalmologic evaluation. The presence of periorbital ecchymosis or subconjunctival hemorrhage should alert the surgeon to the possibility of orbital fracture. The infraorbital nerve is often contused or trapped in its course along the orbital floor, resulting in hypoesthesia in its distribution. Palpation of the orbital rims may elicit pain and reveal step-off deformities. Diplopia and restricted extraocular eye movements can result from entrapment of periorbital soft tissue and/or extraocular muscles. CT examination of pediatric orbital fractures is crucial to both confirm the physical findings and delineate the fracture pattern.

The traditional absolute and relative indications for operative management of orbital floor fractures commonly used in the adult patient do not apply to pediatric patients. The goals of treating pediatric orbital fractures are to restore globe position and correct visually handicapping diplopia. Treatment should begin with pediatric ophthalmologic evaluation and coordination. With nondisplaced or minimally displaced fractures, a conservative approach with careful weekly follow-ups can often yield excellent results. For trauma to produce posttraumatic enophthalmos or vertical orbital dystopia, there must be a composite injury to the orbital bone and supporting structures (ligaments and periosteum). This composite injury allows the intraorbital volume to expand, creating changes in globe position. In our experience, pediatric patients are more likely to receive a traumatic injury that causes bony disruption but that does not lead to enophthalmos,



Fig. 40-8 Left-sided orbital floor trapdoor fracture with trapped muscle.

vertical orbital dystopia, or visually handicapping diplopia.⁴³ This may be a result of greater resiliency of the ocular supporting structures in pediatric patients.

Therefore we recommend that orbital fractures in the absence of acute enophthalmos and vertical orbital dystopia initially be approached conservatively with close surgical and ophthalmologic follow-up, regardless of the bony fracture pattern (Fig. 40-9). With malposition of the globe or persistent diplopia, the goals of surgical treatment should be restoration of orbital volume and release of all trapped soft tissue and muscle to correct globe position and diplopia. Early intervention is indicated when there is significant alteration of orbital form and volume, resulting in vertical orbital dystopia and/or enophthalmos, because delayed repair is often difficult in children—they heal fractures quickly.

Many approaches to the pediatric orbit have been described. A transconjunctival approach is preferred when available, because it has a lower incidence of ectropion and excellent postoperative cosmesis. If extended exposure is needed, a drawback of the transconjunctival approach is the need to perform an additional lateral cantholysis to approach the lateral orbital wall, and this may result in altered morphologic structure of the lateral canthal region. Alternatively, a subciliary or midlid incision can be used. Although the incidence of ectropion with this approach is slightly higher than that of the transconjunctival approach, the cosmesis is quite good in skilled hands. Infrequently an upper gingivobuccal sulcus incision is needed to adequately view and reduce infraorbital rim fractures.

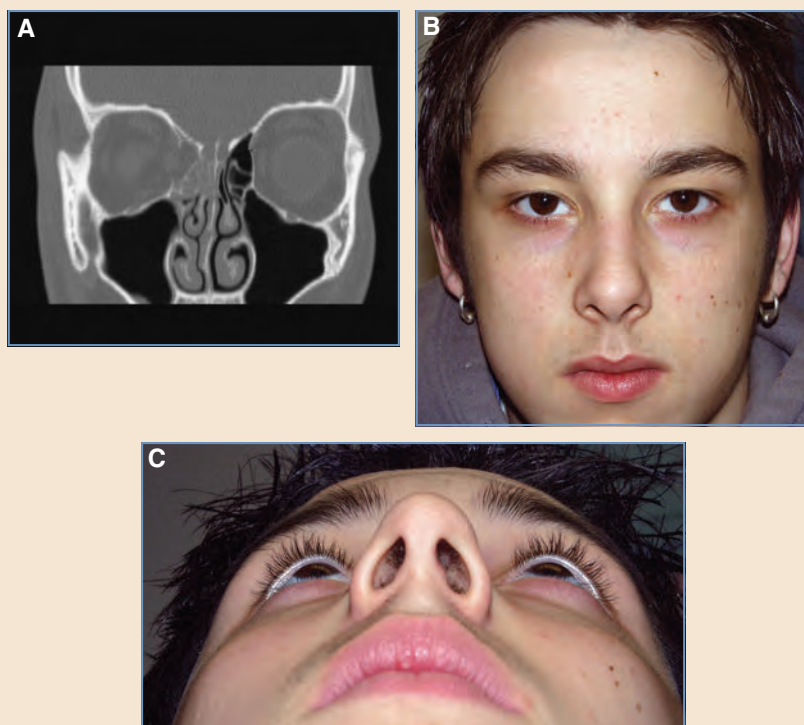


Fig. 40-9 A, Right-sided medial orbital wall fracture treated conservatively. B, Frontal view and C, submental view at 6-month follow-up.



Fig. 40-10 Left-sided orbital reconstruction with split calvarial bone grafts.

When surgically treating orbital fractures, the surgeon must be keenly aware of the differential depth of the orbit to avoid injury to the optic nerve. After adequate mobilization of the orbital contents, the herniated soft tissues are reduced from the fracture defect. The perimeter of the bony defect must be fully delineated so adequate reduction can be ensured, and stable ledges are identified to place grafts. Bony fragments that may impede anatomic restoration are removed. The defect is then repaired with either an autologous split calvarial bone graft or synthetic material, such as resorbable mesh. In the growing craniofacial skeleton, every attempt should be made to use autologous methods of reconstruction. Split calvarial bone grafts or split rib provide excellent bone for orbital reconstruction (Fig. 40-10). Orbital bone grafts should be rigidly fixed, with the smooth cortical surface facing the intraorbital space. Rigid fixation of the orbital rims can be carried out with absorbable microplates. Alternative means of fracture fixation, including metallic microplates and screws or interfragmentary wire fixation, are available but should be used with caution in the growing craniofacial skeleton. Resuspension of the midfacial soft tissues is required to avoid postoperative soft tissue ptosis. Diplopia and ocular movement abnormalities in the immediate postoperative period often resolve, provided there is adequate reduction and release of all entrapped tissue. Monitoring vision often necessitates overnight stays for children undergoing surgical treatment of orbital fractures. Steroids can be given if significant intraorbital surgery is performed. Any ipsilateral eye pain that is unexpected or out of proportion, with or without change in vision, requires thorough examination and possible surgical reexploration.

Zygomaticomaxillary Fractures

Isolated classic zygomatic fractures are uncommon in children. The incidence of zygomatic fractures in pediatric facial trauma is 4.7% when isolated but increases to 16.3% when associated with orbital fractures¹ and are more common with increasing age.²³ This is likely because of the underdevelopment of the maxillary sinus in early childhood, which allows the zygomaticomaxillary buttress to provide rigid support. Significant force is required to cause a zygomatic fracture, because the immature bone is quite resilient. Fracture dislocation occurs through the zygomaticofrontal suture, downwardly displacing the zygoma and orbital floor and often resulting in an oblique craniofacial fracture pattern.¹⁹

The goal of treating pediatric zygomaticomaxillary fractures is restoration of preinjury facial appearance. Two functional deformities may result from significantly displaced zygomaticomaxillary fractures: *orbital dysmorphism* (enophthalmos and vertical orbital dystopia) and *malocclusion*. Nondisplaced or minimally displaced fractures are similarly treated with conservative care and close follow-up. For displaced zygomatic fractures, open reduction internal fixation is required. Surgical exposure is provided by an upper buccal sulcus incision combined with either a subciliary incision or subconjunctival incision with lateral cantholysis to approach the zygomaticofrontal suture.¹⁹ A brow or coronal incision is rarely needed to treat isolated zygomaticomaxillary fractures. Reduction of the fracture is obtained at the lateral orbital rim or zygomaticofrontal suture, the lateral orbital wall or greater wing of the sphenoid, the inferior orbital rim, and the zygomaticomaxillary buttress. The lateral orbital wall or greater sphenoid wing is the key to adequate reduction. After reduction, fixation at the zygomaticofrontal suture, inferior orbital rim, and zygomaticomaxillary buttress is performed, in that order, to adequately stabilize the reduced fracture. The orbital floor should be explored in comminuted zygomatic fractures before and after reduction to ensure that fixation did not change orbital floor stability or orbital volume. Care must be taken to avoid injury to developing tooth buds in young children. Postoperative malalignment of zygomatic fractures has been reported to reach 10%, and this is likely from open “nonreduction” and fixation. Complications are also more common with closed reductions.^{50,51}

Maxillary and Midface Fractures

Isolated midface fractures in children are rare.⁵² This is an effect of the prominence of the cranium and mandible, which generally absorb most of the impact during trauma. Therefore when midface fractures are seen in children, efforts should be made to identify other injuries, because the incidence of associated injuries ranges from 25% to as high as 88%.^{1,19,53,54} Because the classic buttresses are underdeveloped, pure LeFort fractures are very uncommon, and as previously described, children often have oblique patterns of craniofacial fracture.²⁷ The LeFort I fracture is rare, because the maxillary sinus is often not yet pneumatized, and unerupted teeth act as an internal support to the midface. Thus the force is transferred to the alveolus, resulting in tooth avulsion and alveolar fractures.⁵ A midface fracture occurring at the LeFort II level typically presents as a combined nasoorbital ethmoid and LeFort I maxillary fracture and is often unilateral. Midface fractures occurring at the LeFort III level are very uncommon in children and classically present as combined nasoorbital ethmoid and zygomaticomaxillary fractures. Bony injuries at this level present as multipiece midface fractures. In contrast, because the palatal suture is incompletely ossified, midline palatal fracture separations are common in children.¹⁹ The diagnosis of a midface fracture is made by the physical findings of maxillary mobility, bony step-offs, and malocclusion. Fine-cut CT scans should be used to delineate the injury and prepare for surgery. Intracranial injury must be ruled out before surgical intervention begins, because significant force is required to produce midface fractures in children.

Typically, a conservative approach should be taken with infants and young children who have minimally displaced or greenstick maxillary fractures, and this usually includes a liquid or soft diet. A conservative approach is possible because pediatric patients have rapid healing and the ability to remodel their fractures, and it allows the option of early orthodontic intervention to correct mild occlusal discrepancies. In older children and adolescents, a more classic approach should be used. If maxillomandibular fixation (MMF) is required, care must be taken to avoid injuring the developing tooth buds and subluxating or extracting primary teeth with conical roots. MMF can be difficult in infants and young children, and the surgeon must be creative to achieve rigid intermaxillary fixation (IMF). The options of circummandibular wiring and piriform aperture

drop wires can be used to secure fixation (Fig. 40-11). In addition, alternative techniques, such as “homemade IMF hooks,” (Fig. 40-12) can be fashioned as needed in the dental laboratory, tailored to the patient, and used with considerable ease and effectiveness. Also, if the dental units are present, arch bars can be placed during the period of primary or mixed dentition.⁵⁵ Proper occlusion must be obtained before fixation and checked periodically. Palatal fractures can often be treated with closed reduction and fixation with an orthodontically made palatal splint without rigid fixation. Cooperative interaction with pediatric dentists and orthodontists can often

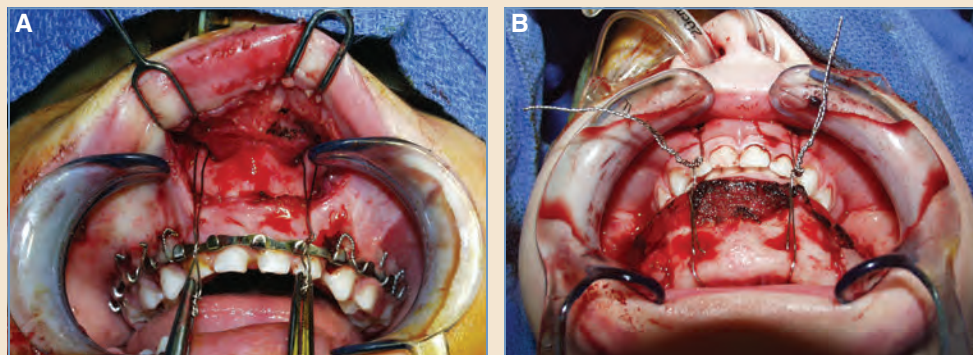


Fig. 40-11 A, Piriform suspension wires used for maxillomandibular fixation. B, Mandibular border wires, similar to circummandibular wires, used for maxillomandibular fixation.

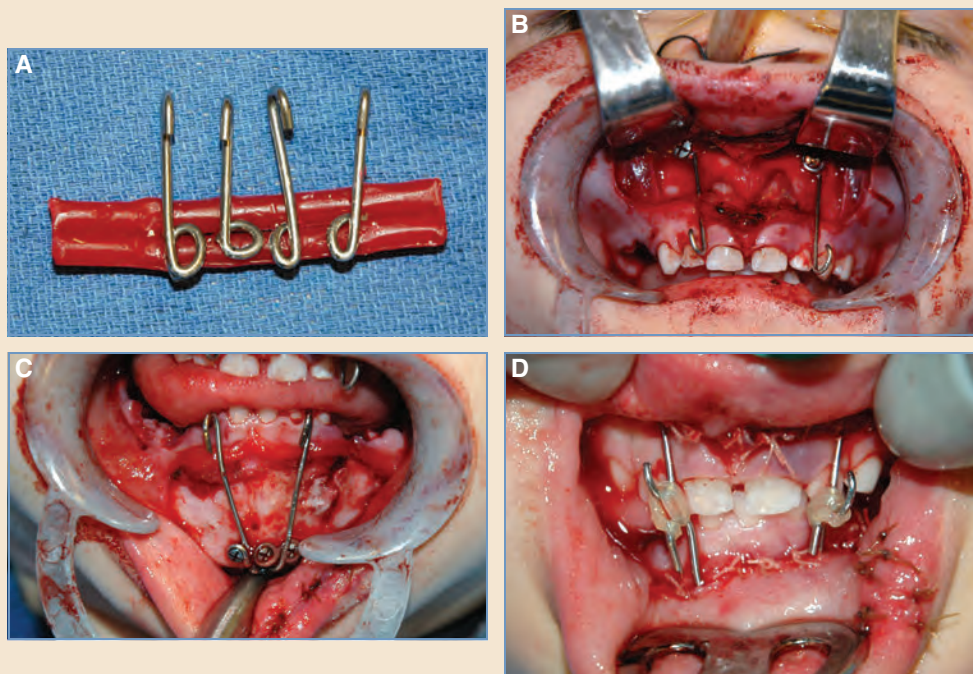


Fig. 40-12 A, “Homemade” IMF hooks. B, IMF hooks secured to the piriform region of the maxilla in a mixed dentition patient. C, IMF hooks secured to the inferior mandibular border. D, After closure with elastics in place.

save a significant amount of effort, especially if preoperative impressions and models were taken before injury and are available. In infants and children, the length of MMF is often less than the 6 weeks used for adult patients. After a shortened course of rigid MMF, dental elastics are used to maintain the desired occlusal relationship. Because there is significant risk of growth disturbance regardless of adequate treatment, parents of children with midface fractures must be informed of potential long-term growth effects and encouraged to follow up closely.

Nasal Fractures

Nasal fractures are the second most common facial fracture in children and can often involve the nasal bones and cartilages and the septum. Because of the significant presence of cartilage, a nasal fracture is often missed during examination in pediatric patients.⁵⁶ In children, the nasal bones can separate along an open midline suture, thus predisposing them to an “open book” type of fracture, which results in the nasal bones overriding the frontal process of the maxilla.¹⁰ Pediatric nasal fractures may have a significant impact on the growth and development of the facial skeleton, resulting in septal or nasal deviation, as well as midface hypoplasia. Even with early intervention and adequate reduction, cosmetic and functional deformities, such as deviations, dorsal humps, saddlenose deformities, and airway obstruction, still occur. Therefore the surgeon should discuss with parents the potential complications and the need for further corrective surgeries in the future. Unless significant symptomatic nasal obstruction is present, secondary nasal surgery should be performed after skeletal maturity has been reached.

It is often necessary for children to be under general anesthesia for adequate nasal examination and treatment. A complete intranasal examination is routinely required to thoroughly evaluate the septum. As with other pediatric facial fractures, plain radiographic films are grossly inadequate to diagnose nasal fractures in pediatric patients. Fine-cut CT scans provide the necessary information for complete diagnosis and treatment. It is prudent to treat open fractures and septal hematomas acutely and without delay, whereas other types of nasal fractures often require a short delay to allow swelling to decrease and reveal potential deformities. One unique injury found in pediatric nasal trauma is a hematoma between the upper lateral cartilages and nasal bones.^{19,56} Because these two structures are overlapped and loosely attached, the cartilages can easily detach, resulting in a hematoma between the cartilage and bone. This injury requires drainage through an intercartilaginous or subperiosteal incision.⁵⁶

An untreated septal hematoma can lead to pressure necrosis and diminished blood supply to the septal cartilage. This can result in either thickening or destruction of the septum and classic saddlenose deformity. Adequate drainage of a septal hematoma requires an incision through the mucoperichondrium on the side of the hematoma at the most dependent portion. When present bilaterally, bilateral mucoperichondrial incisions or resection of a small piece of the inferior septum through a unilateral incision can be used for drainage. Silicone intranasal splints and/or nasal packing can be used for postoperative septal compression during the healing phase.

Acute treatment of nasal fractures with closed reduction and external fixation often leads to suboptimal results in adults.⁵⁷ This is most likely caused by inadequate release of tension forces deviating the septum, nasal cartilages, and bony pyramid when closed reduction is used alone. Adequate treatment of a crooked nose usually requires a complete septorhinoplasty, including correcting any septal deviation and releasing deforming forces. However, aggressive nasal surgery in infants and children is still thought to have a negative impact on the growing facial skeleton. Because of this concern, we think that it is most appropriate to attempt closed reduction and external fixation of nasal fractures in the acute setting when there is obvious nasal bone deformity. This reduction of the nasal bones and septum should be attempted with the patient under sedation or general anesthesia. An Ash forceps is used to reduce and relocate the septum. A knife

handle or Boies elevator, placed endonasally and underneath the nasal bones, is combined with external digital pressure to reduce the nasal bones. Intranasal packing is often required to maintain the reduction and is used in conjunction with an external nasal splint. When intranasal packings are used, antibiotics should be given to avoid the potential risk of septic shock syndrome.⁵⁸ Because facial fractures heal quickly in younger children, splints are rarely needed after 5 days.

Mandibular Fractures

Mandibular fractures are the most common facial fractures reported in children, and this is likely because the bony mandible is prominent. The reported incidence of mandible fractures is between 20% and 40% of all pediatric facial fractures.^{21,59} This high incidence may also be caused by the probable underreporting of nasal and dentoalveolar fractures. Mandibular fractures are uncommon in children younger than 6 years, and therefore their presence should alert the physician to look for concomitant injuries. The incidence of pediatric mandibular fractures increases between 6 and 15 years of age.³ As children grow, the distribution of fractures in the various regions of the mandible also changes. Condylar fractures are common during the first 6 years of life (43.4%), but this decreases to 7% to 10% in children ages 13 to 18 years.^{5,60} In children older than 15 years, 76% of mandibular fractures occur in the regions of the angle and body.⁶⁰ The characteristics of pediatric mandibular fractures also differ from those in adults and include more greenstick fractures and long irregular sagittal fractures.⁶¹ All of these issues should be taken into consideration before the management of pediatric mandibular fractures is undertaken. Attention should be given to cervical spine injuries, because the force required to produce a fracture is significant.

The most common symptoms of pediatric mandibular fracture are pain and malocclusion. The patient often complains that his or her teeth do not come together. Drooling and trismus, with a decreased maximal incisal opening, are often present. A dental step-off, with bleeding, swelling, and ecchymosis, is routinely found intraorally (see Fig. 40-2). In addition, classic patterns of malocclusion are easily noted. Although bilateral condylar fractures usually result in an anterior open bite, unilateral condylar fractures often present with contralateral posterior open bites. With condylar neck fractures, the condylar head is often medially displaced from the pull of the pterygoid muscles. The TMJ can be assessed by placing fingers in the external auditory canal while the child opens and closes his or her mouth. Another useful technique to diagnose pediatric mandible fracture is simply to place the heel of the examiner's hand on the child's symphysis and ask the child to push against it with his or her chin. If there is significant pain, then the suspicion of mandible fracture should be raised. Although a Panorex is useful in older children, a CT scan is the most reliable way to delineate mandibular fractures in the pediatric population.

To safely treat children who have mandibular fractures, one must understand basic dental development. The deciduous mandibular incisors first appear at approximately 6 months of age, followed by eruption of the molars and then the cuspids. Even with their conical shape, primary tooth roots are strong enough for interdental wiring between 3 and 6 years of age, but care should be taken when circumdental wires are placed.⁵⁵ Mixed dentition is present between 6 and 12 years of age, and the resorption of primary tooth roots, along with the underdevelopment of permanent tooth roots, creates a unique challenge for securing arch bars in this age group. Therefore it is helpful to perform a Panorex study with these patients to delineate teeth with stable roots. The surgeon must be creative and well versed in all manner of interdental fixation, such as acrylic splints, circummandibular wiring, drop piriform aperture suspension wiring, and nasal spine suspension wiring. Although teeth are available for placement of circumdental wires and arch bars in younger patients (3 to 4 years of age), their inability to understand and cooperate can often result in disruption of IMF in the early postoperative period because of their constant struggle against



Fig. 40-13 A, Child shown in a cervical collar. B, Child shown in an Ace wrap bandage.

fixation. In this younger age group, we suggest that circummandibular and piriform suspension wires, in addition to arch bars, be placed for added maintenance of IMF.⁵⁵

When the surgeon is treating jaw fractures, occlusal relationships are of the utmost importance. If available, every effort should be made to obtain up-to-date preinjury models to make occlusal splints. Alternatively, preoperative impressions can be taken of the child's injured jaws, and these impressions can be used to create stone models that may be cut, manipulated, and mounted on an articulator. After the ideal occlusion is established with the models, a surgical splint is made and used intraoperatively to guide the surgeon.

Treatment of pediatric mandibular fractures should be tailored to address each region of the mandible specifically. The goals are the same as for adults: restore normal occlusion, ensure bony union, and avoid infection.¹⁹ Because the pediatric mandible is full of developing dental follicles, a conservative approach to the treatment of fractures is always indicated.⁵⁹ Minor malocclusions from fractures are often well tolerated in infants and young children, because children have a remarkable ability to remodel their fractures well, and an orthodontist may manipulate and correct minor occlusal discrepancies at a later date. Therefore, if a fracture creates minimal displacement and the occlusion is essentially normal, a conservative approach is warranted. This usually includes a soft diet, dental hygiene, and mandibular rest. Some type of external splint, such as a cervical collar or an Ace wrap applied in the craniocaudal direction, can also be used (Fig. 40-13).

For patients with displaced fractures and significant malocclusion, surgical intervention is indicated. The specific surgical intervention depends on the fracture pattern. For single fractures (subcondylar) in infants and young children, closed manipulation should be performed first, and if normal occlusion is obtained, a course of MMF or functional elastic therapy can be used.⁶² If combination fractures are present (subcondylar and parasymphiseal) or if normal occlusion cannot be obtained, then open reduction and internal fixation is indicated. Internal fixation in infants and young children is a topic of controversy and should be minimized if at all possible. Usually an inferior border wire is preferable to metallic plate fixation (Fig. 40-14); however, resorbable miniplate fixation has gained favor when internal fixation is warranted. As discussed previously, arch bars often can be challenging to place in a pediatric patient, and alternative forms of MMF can be used (see Fig. 40-12). For older children who no longer have mixed dentition, routine

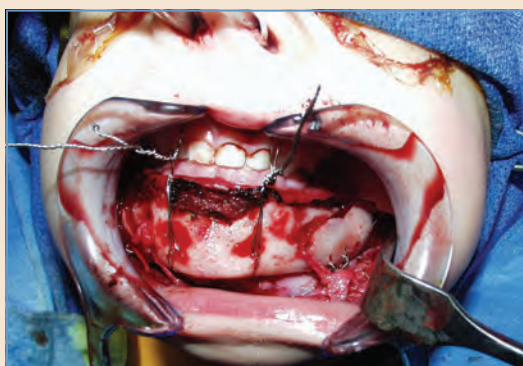


Fig. 40-14 Left-sided inferior mandibular border wire fixation of left-sided sagittal body fracture.

principles and techniques of mandibular fracture treatment apply. Many surgeons consider it appropriate to remove all nonresorbable fixation after fracture healing in growing children.

Condylar Fractures

The condylar head is considered a growth center of the pediatric mandible, and in infants and young children they are nothing more than “vascular bony sponges.”⁶³ Any injury that interrupts the blood supply and alters the morphologic structure can easily result in TMJ ankylosis and subsequent alteration of mandibular growth. The vast majority of condylar head or neck fractures in infants and young children should be treated conservatively; however, the specific treatment for a condylar fracture depends on the location of injury. Intracapsular condylar head injuries should be treated with a soft diet, gentle mobilization, and physical therapy, because the risk of ankylosis is significant with immobilization. When condylar head injuries occur in combination with other mandibular fractures, it may be an indication for open reduction and internal fixation of the other fractures so that immediate range-of-motion therapy and physical therapy may be started. Chewing gum can be a powerful tool for TMJ mobilization, along with other devices, such as a TheraBite (Atos Medical AB) to encourage mouth opening. The indications for opening a child's TMJ are few, and they include a foreign body in the joint or an inability to range the joint into occlusion. A displaced condyle in the middle cranial fossa is an extremely rare occurrence.³

Subcondylar Fractures

Most condylar neck fractures in infants and young children are treated with closed reduction and a short course of functional elastic therapy. Arch bars can be placed, and often a single contralateral elastic is all that is required. Subcondylar fractures in children are frequently greenstick, incomplete fractures that remodel well when treated conservatively with a soft diet and careful monitoring. If injuries are bilateral or there is a loss of ramus height resulting in an anterior open bite, closed reduction and external fixation with a short course of MMF (2 to 3 weeks) may be indicated. In adolescents, bilateral condylar neck fractures resulting in an anterior open bite may be an indication for open reduction and internal fixation of the condylar neck in the hands of those comfortable with that approach.

Body and Angle Fractures

When body and angle fractures are significantly displaced and/or associated with condylar head or neck fractures, open reduction and internal fixation (ORIF) is indicated. Often these fractures are sagittal and longitudinal. Whenever possible, intraoral incisions should be used for open treatment of pediatric mandible fractures. A transbuccal approach can be challenging and may



Fig. 40-15 A, Bilateral mandibular fracture in a 3-year-old boy. B, Axial CT scan showing bilateral sagittal mandible body fractures. C, Coronal CT scan of left condylar head fracture. D, Immediate postoperative axial CT scan of reduced mandibular body fractures with bilateral inferior border wire fixation. E, Six-month postoperative three-dimensional CT scan demonstrating normal mandibular morphologic structure. F, One-year follow-up with normal age-appropriate occlusion. G, One-year follow-up with normal maximal incisor opening.

result in inadequate reduction. When the surgeon is approaching the symphyseal region, care must be taken to leave a cuff of mentalis muscle on the mandible for resuspension to avoid the postoperative “witch’s chin” deformity. The mental nerve must be visualized to avoid injury and postoperative anesthesia. During open reduction, soft callous is gently curetted from the fracture site; the surgeon must be aware of developing tooth follicles and nerves.

Treatment of symphyseal or parasymphyseal fractures may require wiring of the mandibular border and dental bridle wires to adequately reduce and fix the fracture (Fig. 40-15). This fixa-



Fig. 40-16 Inferior mandibular border miniplate fixation.

tion technique usually treats the fracture adequately if a short course of MMF is also used, and it theoretically poses fewer disturbances to subsequent growth and development. If required or if MMF cannot be used, further fixation can be obtained with an inferiorly placed resorbable or metallic miniplate with screws placed monocortically and “aimed” away from developing tooth buds (Fig. 40-16). If metallic plates and screws are used, it is often recommended that they be removed after there is evidence of bony healing to prevent potential growth disturbances.⁵⁹

Alveolar Fractures

Treatment of alveolar fractures requires reduction of the dentoalveolar segment, fixation with wires or arch bars, and/or dental splinting with acrylic splints. This provides stabilization with little to no discomfort and, for older and cooperative children, can occasionally be done in an office setting. Postoperatively, dental hygiene and a soft diet are prescribed. Any minor occlusal malalignment can be addressed orthodontically after the fracture is completely healed. Close dental follow-up is encouraged.

Panfacial Fractures

Panfacial fractures, by definition, involve the upper, middle, and lower craniofacial skeleton. This fracture pattern is exceedingly rare in children. When children have panfacial fractures, they are usually associated with multiorgan injuries that make the management of these fractures challenging. Multiple schools of thought regarding the sequence of fracture repair have been proposed by different authors, and multiple approaches are often used to address the frontal region, upper midface, lower midface, occlusion, and basal mandibular area.⁶⁴ These include coronal, lower eyelid/transconjunctival, upper gingivobuccal sulcus, lower gingivobuccal sulcus, and preauricular-retromandibular incisions. The general goal is to reduce facial fractures to the skull base with an “outside-in” approach. This includes reducing the naso-orbital-ethmoid fracture to the nasion, the mandibular fracture to the glenoid fossa and TMJ, the zygomatic arch fracture to the temporal bone, and the zygomaticomaxillary complex fracture to the greater wing of the sphenoid and at the zygomaticofrontal suture. Our preferred sequence for pediatric panfacial fractures starts with direct reduction and fixation of palatal fractures and establishment of

an anatomic upper dental arch. This is followed by addressing subcondylar fractures, which establishes posterior mandibular height and reduces the mandible to the glenoid fossa-TMJ-skull base. After this, MMF is established and mandibular body fractures are fixated to establish lower facial width and occlusion. Next, the zygomaticomaxillary fractures are addressed. The zygomatic arch is reduced to the temporal skull base to establish anteroposterior projection of the face. The zygoma is reduced to the greater wing of the sphenoid-skull base and at the zygomaticofrontal suture. The nasoorbital-ethmoid fracture and central fragment are then reduced to the nasion. Reduction and fixation of the inferior orbital rim and LeFort buttresses follow.

Finally, orbital reconstructions are performed, which complete the panfacial fracture. It is important to develop a comprehensive treatment plan before surgery. This often includes coordination with the neurosurgery department for transcranial access, obtaining preoperative models for fabricating occlusal splints, and arranging with other surgical specialties and pediatric anesthesia to maximize the resuscitation of the patient before the prolonged surgery he or she may need to endure.

POSTOPERATIVE CARE

The immediate postoperative period may be exceedingly stressful for pediatric patients who undergo surgical treatment of facial fractures. This is especially true when IMF is involved. Parental presence and constant reassurance are essential parts of postoperative care. Surgically, two areas need special attention. First, the airway needs to be protected. This requires experience in the care of these patients to know when a surgical airway is required, and the decision should be made before fixation of the fractures. Second, in patients with fractures involving the orbit, thorough serial ophthalmologic examinations should be performed. Any signs of superior orbital fissure syndrome or orbital apex syndrome must be addressed immediately.

Antibiotics are not essential but should be used when specifically indicated. When bone grafts or hardware are used during the fixation of facial fractures, we advocate a short 3- to 5-day course of antibiotics. Antibiotics are also indicated when there is salivary contamination of the fractures or when the frontal sinus and intracranial space are involved. Careful wound care and oral hygiene are also very important. Our protocol includes antibiotic mouthwash every 4 hours in the immediate postoperative period and antibiotic ointment on all skin incisions.

The role of parenteral steroids remains controversial. Their use is considered most beneficial to decrease swelling in the airway and posterior orbit. A single intravenous dose of 0.5 to 1.0 mg/kg of dexamethasone is usually given.⁶⁵ Repeating and/or tapering additional doses is often considered. Higher doses may be required for optic neuropathy, and the guidance of a pediatric ophthalmologist is recommended. The potential risks associated with steroid use include postoperative infection, aseptic necrosis of the hip, and adrenal insufficiency, all of which are rare.

Routine postoperative CT scans are not always indicated; however, they are invaluable to evaluate reduction and fracture alignment and to document postsurgical treatment. In our opinion, these studies remain the most valuable learning tool for surgeons who have limited experience with surgical management of pediatric facial fractures. All children who have sustained facial fractures require annual follow-up during the period of growth and development of their craniofacial skeleton. Annual physical examination and assessment by a craniofacial surgeon and orthodontist are suggested. Yearly cephalograms and Panorex radiographs, along with dental impressions and standardized photographs, are obtained to document and track facial growth and development. We cannot stress enough the importance of serial dental and facial photographs taken at standard settings. These annual photographs not only document the injury and wound healing but also show subtle soft tissue, skeletal, and dental changes that may or may not occur after fixation of facial fractures.

A coordinated team approach is recommended for the care of pediatric facial fracture patients, similar to the care for cleft and craniofacial patients. The team should include the craniofacial surgeon, pediatric neurosurgeons, pediatric orthodontists and dentists, a pediatric ophthalmologist, a social worker, and a pediatric developmental specialist/psychologist who will focus on the developmental and psychological impact of the injury.

POSTOPERATIVE COMPLICATIONS

Postoperative complications after the treatment of pediatric facial fractures can be divided into three categories: those that occur as a result of the injury, those that occur as a result of the treatment, and those that occur over time with altered growth and development.^{66,67} The most drastic immediate complication is compromise of vision. As previously noted, even though children suffer fewer facial fractures in general, when they do, there is a higher incidence of blindness compared with adults. It is imperative to have a pediatric ophthalmologist evaluate the patient preoperatively to document a baseline examination and diagnose any injury. A postoperative examination should also be completed. If vision deteriorates in the immediate postoperative period, timely surgical reexploration is warranted.

Infection can also occur in the immediate postoperative period but is very unlikely in healthy young patients. Infection is more concerning when hardware or bone grafts were used for reconstruction. Any early infection requires judicious use of culture-specific antibiotics, drainage, and irrigation. Infections in the oral cavity after open reduction and internal fixation with hardware exposure can often be treated with antibiotics and good oral hygiene without hardware removal. True infections elsewhere in the craniofacial skeleton are more concerning and may require washout and removal of hardware. Bone grafts rarely become infected, but if this occurs, the graft invariably requires removal. This usually creates significant defects that are cosmetically and functionally displeasing when the soft tissue envelope collapses and scars. These patients frequently require extensive secondary reconstruction with autologous split calvarial or rib grafts. It is prudent to perform a delayed reconstruction, allowing the infection to subside and avoiding further graft loss. Infection can also occur in the subacute setting, which is more common when

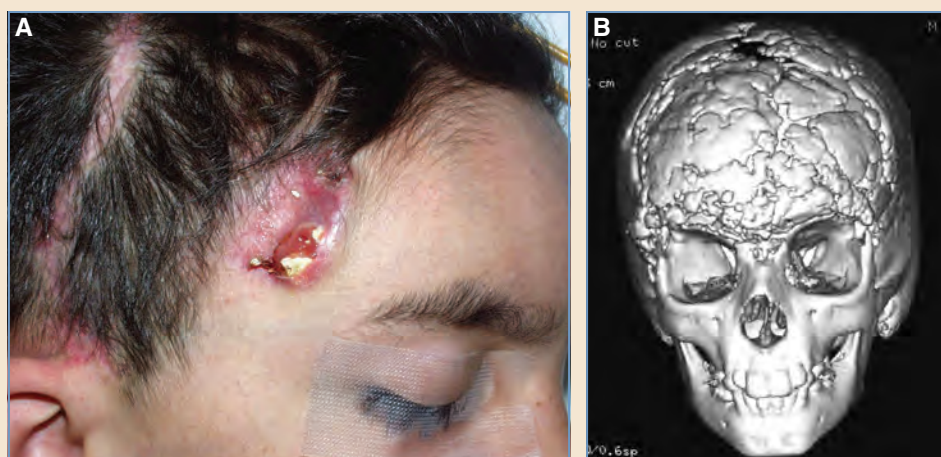


Fig. 40-17 A, An infected bone substitute extruding through the temporal wound. B, A three-dimensional CT scan showing the fractured, infected, and extruding bone substitute.

nonautologous materials are employed, such as those used for cranioplasty (Fig. 40-17). When these infections occur, all of the offending material must be removed, and a delayed secondary autologous reconstruction must be performed.

Late postoperative complications in children with craniofacial fractures include bony contour deformities (for example, saddlenose), ophthalmologic disturbances (for example, enophthalmos and orbital dystopia), orthognathic deformities (for example, malocclusion) (Fig. 40-18), and the rare “growing skull fracture.” It is not uncommon to have minor contour discrepancies after the initial treatment of craniofacial fractures when unpredictable growth and development follow. These deformities most often do not require significant surgical corrections. However, when major bony deformities develop, extensive reconstruction with autologous calvarial bone or rib grafts is usually required.

Postoperative ophthalmologic disturbances include orbital volume-related problems such as enophthalmos and orbital dystopia, as well as soft tissue and scarring problems, including ectropion and extraocular muscle dysfunction. The goal of reconstruction is to restore pretraumatic orbital volume and morphologic structure. When this restoration is inadequate or when bone grafts are absorbed and soft tissues atrophy, enophthalmos may occur. Characteristic areas of bony loss include the orbital floor and posteromedial and lateral orbital walls. The basis of secondary orbital volume restoration is purely anatomic. This begins with alignment of the lateral orbital wall (greater wing of the sphenoid), establishing the key landmark for reduction of all other components of the orbital fracture. Frequently the malaligned zygomaticomaxillary fracture must be reosteotomized to mobilize and reduce the fracture segment. Additional osteotomies may be required to restore natural anatomy. After these units are stabilized by miniplates, the floor and medial wall defects can be repaired. A coronal incision may be needed to approach the posterior aspect of the medial wall.

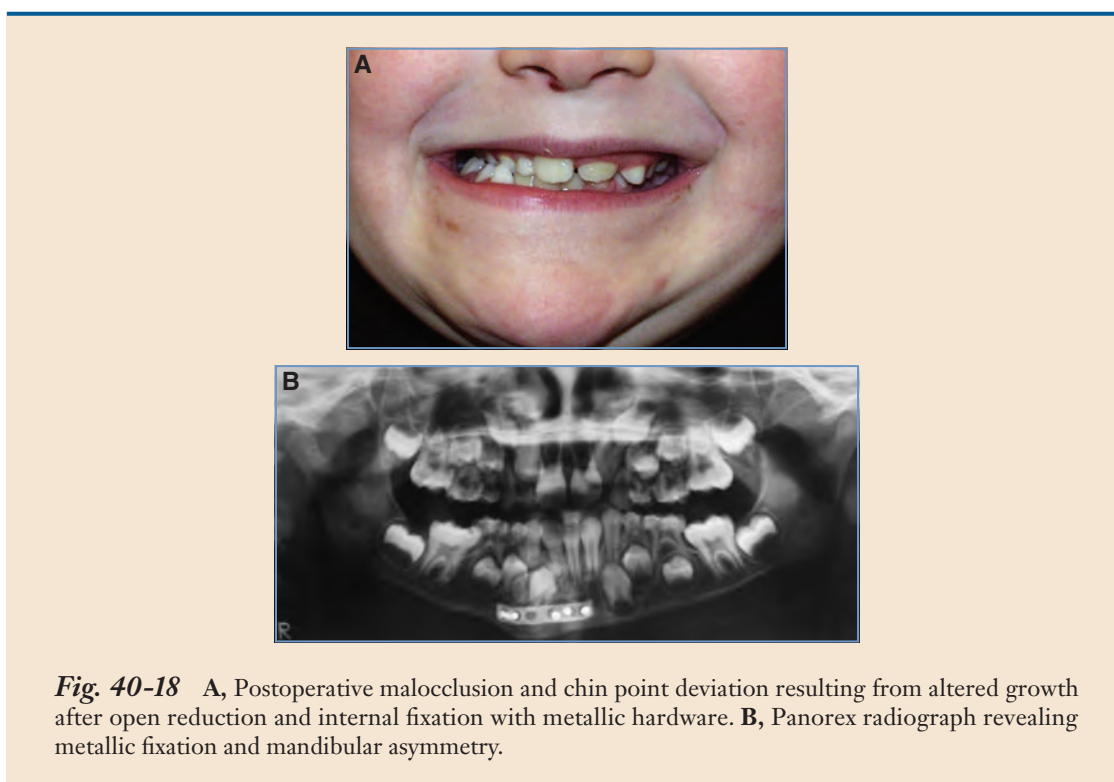


Fig. 40-18 A, Postoperative malocclusion and chin point deviation resulting from altered growth after open reduction and internal fixation with metallic hardware. B, Panorex radiograph revealing metallic fixation and mandibular asymmetry.

Posttraumatic exophthalmos rarely occurs. It is most likely caused by a superior orbital roof fracture and herniation of the brain into the bony orbit. This generally requires a coronal incision and frontal craniotomy to reduce the herniated brain and place a bone graft and pericranial flap to cover the defect. Exophthalmos can also occur with excess correction and reduction of orbital volume. In pediatric patients, a growing skull fracture can also occur after an orbital roof fracture, and this may cause pulsatile exophthalmos.

Secondary orthognathic deformities occur in recognized patterns after facial fracture with or without treatment.⁶⁸ An elongated face with an anterior open bite may occur after LeFort fractures that are inadequately reduced and the posterior maxilla is inferiorly and posteriorly displaced.¹ This pattern of malocclusion can also occur when posterior mandibular height is lost by improper fixation of mandibular fractures or when the condyles are not correctly seated into the fossa at the time of the repair. To correct an anterior open bite, the fracture pattern is re-created, and anatomic reduction is performed with IMF. A posterior maxillary crossbite can occur if the ipsilateral mandibular ramus or angle fracture is inadequately reduced. This often requires orthodontic treatment to correct and, if severe enough, secondary orthognathic surgery.

As previously discussed, the condyles in the mandibles of pediatric patients are growth centers. Therefore condylar injuries have the potential to disrupt bony growth, causing asymmetry and malocclusion. This most commonly presents as secondary asymmetrical mandibular retrognathism and a decreased maximal incisal opening, giving the patient an ipsilateral occlusal cant and a deviated dental midline. To correct this deformity, distraction osteogenesis can be performed; alternatively, orthognathic surgery can be done after skeletal growth has been completed. The typical orthognathic procedures used to correct this deformity are bilateral sagittal split osteotomies of the mandible with a possible combined LeFort I osteotomy with or without osseous genioplasty.

LONG-TERM RESULTS

Despite timely and adequate treatment of pediatric facial fractures, long-term results remain unpredictable. Growth and developmental disturbances may occur from (1) the fracture itself, (2) an open approach and periosteal undermining, (3) rigid fixation of the fracture, and (4) disruption of growth centers within the pediatric facial skeleton. Numerous reports have documented unpredictable growth patterns and delayed deformities in the pediatric population after facial trauma. For example, growth disturbances associated with nasal fractures occur from a suspected premature ossification of the septovomerine suture,² whereas some reports indicate that zygomatic fractures rarely incur any long-term consequences.¹⁰ Orbital fractures occurring before the age of 7 years have been noted to cause growth disturbances; however, orbital growth is often complete after 7 years of age, and fractures after this are rarely associated with abnormal development.¹⁰ Nasoorbital-ethmoid fractures are not common in children; however, when they occur, the disruption of facial growth centers can result in midface hypoplasia in both the vertical and anteroposterior directions.²

The literature lacks comprehensive long-term follow-up of pediatric patients with facial fractures. We are currently involved in establishing a multicenter pediatric facial trauma registry to document the long-term consequences of facial fractures on the pediatric population and have recently published our early follow-up of a cohort of 177 patients over 1 year.⁶⁷ In this study, we introduced a classification system for facial fracture–related adverse outcomes: type 1 adverse outcomes were directly related to the fracture; type 2 adverse outcomes were directly related to the treatment; and type 3 adverse outcomes were related to the interaction between the fracture, treatment, and patient growth. Factors associated with worse outcomes included multiple fractures and fractures that required operative intervention.

KEY POINTS

- Children are not small adults; in addition, 5-year-old patients are not treated the same as 10-year-old patients, and they are not treated the same as 15-year-old patients. Each pediatric patient and each fracture are treated uniquely, applying the principles discussed in this chapter.
- Whenever possible, consideration should be given to treating young patients conservatively.
- CT scans are necessary for adequate evaluation of pediatric patients with facial trauma.
- Whenever possible, absorbable hardware should be used to treat pediatric facial fractures. Caution should be exercised when the surgeon considers the use of nonautologous materials for the growing pediatric craniofacial skeleton.
- When treating pediatric patients with craniomaxillofacial fractures, the practitioner must educate the family on the unpredictable growth and development that may follow these injuries, and he or she should commit to the long-term, adequate follow-up that these patients require.

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Part III



Congenital and Acquired Deformities of the Hands and Upper and Lower Extremities

41

Congenital Hand Anomalies: Embryology and Classification

Aaron Corde Mason • Pundrique R. Sharma



The hand is a complex organ. It is integral to human social interactions, communication, and multiple daily functions. Because of this, developmental anomalies of the hand may significantly affect the life of a newborn infant. Parents are justifiably concerned and seek restoration of normal hand function for their children. Hand anomalies pose significant challenges for reconstructive surgeons. Although aesthetic restoration is ideal, the reestablishment of anatomic foundations for normal hand function in a timely manner is imperative to minimize delays in acquiring gross and fine motor skills during newborn development.

The various causes of congenital upper limb anomalies are beginning to be more fully understood. Many have a genetic basis, although environmental influences can lead to developmental miscues. Congenital anomalies of the upper limb affect 1 in approximately 600 live births.¹ Many anomalies are isolated, but a few are syndromically associated with other systemic anomalies.

The development of the upper extremity is complex, and multiple opportunities exist for genetic and environmental influences to alter its normal development. This has resulted in a host of identified anomalies that have been categorized based on their morphologic features. This gave rise to a classification system that had little to do with actual embryology. As our understanding of normal embryology advanced, new concepts were discovered and new classification systems arose. The intent of this chapter is to provide an overview of normal upper extremity and hand development and to introduce the congenital hand classification systems most used in practice.

UPPER EXTREMITY EMBRYOLOGY

In humans, evidence of upper limb development appears at 26 to 28 days' gestation with the emergence of the upper limb buds.^{1,2} Digits can be first recognized on days 41 to 43, and the process culminates by days 51 to 53 with five segregated digits. Developmental activities are genetically orchestrated during this period. Spatial coordination of activities in a three-dimensional fashion is important for normal limb development, and numerous signals control the organization of tissues along each of the three axes in a process known as *pattern formation*.^{3,4} The three axes are the proximodistal axis, the AP (craniocaudal) axis, and the dorsoventral axis (Fig. 41-1). Each is associated with specialized tissues and tissue factors that direct development in a specific fashion.

The Three Axes

Upper extremity development begins with the expression of *sonic hedgehog* (*SHH*) from the notochord,⁵ leading to the formation of a limb bud from locally proliferating mesodermal cells at the appropriate upper flank level (Fig. 41-2). The limb bud soon evolves with cartilage precursor cells in the center (the chondrogenic core), and other connective tissue cells (destined to be tendons and muscles) accrue peripherally. This aggregate of mesodermal cells leads to a bulging of the ectoderm at the junction of the ventral and dorsal ectoderm, known as the *apical ectodermal ridge* (AER). The cellular activity signals the coalescence of the AER from local ectoderm along the rim of the limb bud. The AER navigates the evolving limb along the proximodistal axis and is responsible for separating the webbed hand through interdigital apoptosis.³ Highly mitotic cells within the mesoderm beneath the AER, in an area known as the *progress zone*, contribute to the growth of the bud laterally and distally. As limb growth progresses distally, more proximal cells leave the progress zone for their final positions within the developing limb.⁶ Cellular differentiation occurs, with more proximal structures evolving toward humeral precursors, followed by radial, ulnar, carpal, and phalangeal precursors distally. At this stage, failure or loss of the AER leads to transverse growth arrest.

Members of the *fibroblast growth factor* (*FGF*) family are also involved in the development of the upper limb.^{1,5-7} *FGF10*, detectable in flank ectoderm before the AER evolves, is a prospective trigger for initial limb bud formation. The AER itself expresses three *FGFs* (*FGF2*, *FGF4*, and *FGF8*) that probably participate in the proximodistal development of the upper limb. Experimentally, the combination of *FGF1*, *FGF2*, and *FGF4* leads to the development of a normal limb, and their application ectopically leads to ectopic limb formation.⁸ The zone of polarizing activity (ZPA) controls the pattern formation of the evolving upper limb in the craniocaudal (radioulnar) axis.^{4,5,7,9} Located on the caudal (ulnar) margin of the growing limb bud, this region contains highly specialized mesenchyme that expresses the protein *SHH*, which is released in a graded fashion along the AP axis of the developing limb. The result is a gradient of *SHH*, which is higher on the caudal edge of the developing limb and causes ulnarization and absent in the cranial edge, allowing radial digits to form.^{7,9} In this model, cells "sense" their location along a gradient, as well as their future positional information. Another model, Turing's reaction-diffusion model, postulates that positional information is not solely the result of a given morphogen's concentration; it is also the result of interactions of different morphogens that can be mathematically described and may be of particular relevance to digital differentiation and development.^{7,10,11}

Not unexpectedly, the AER and ZPA are intricately codependent in the embryogenesis of the upper limb.^{3,4,6} The integrity of the AER is essential to maintain an operational ZPA. *FGF4* produced by the AER supports the ZPA and maintains its expression of the *SHH* protein, which

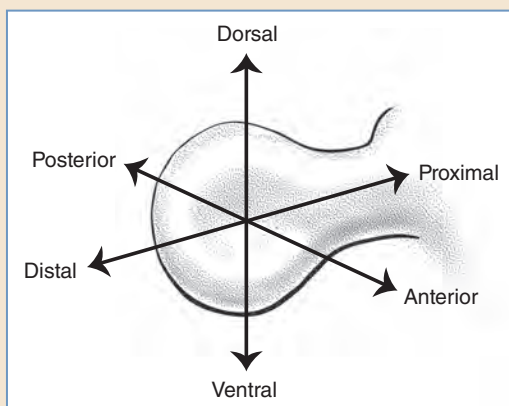


Fig. 41-1 The axes of tissue organization. An arm bud shows the three axes whose differential development leads to the evolution of the human hand.

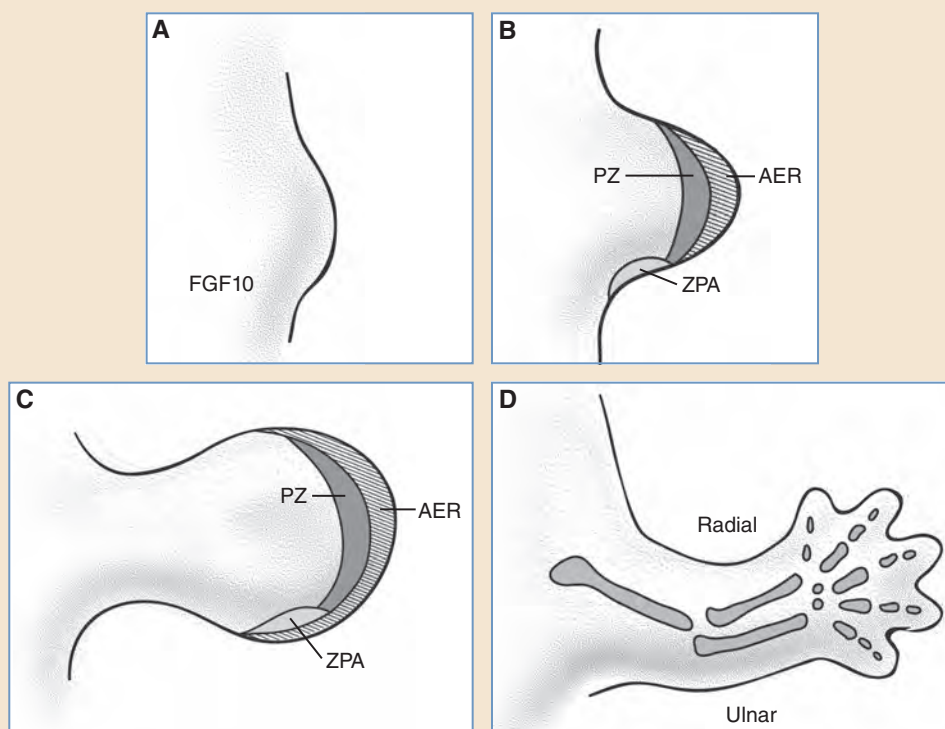


Fig. 41-2 Hand development. **A**, In part because of the influence of *FGF10*, upper extremity development is first noted by the formation of a limb bud from local mesodermal cells on the lateral side of the embryo. **B**, The emergence of the apical ectodermal ridge (*AER*) guides development in a proximo-distal axis. The progress zone (*PZ*) is identified behind the *AER*. The localized zone of polarizing activity (*ZPA*) forms, guiding development of the limb in the AP or radioulnar axis. **C**, As development continues, the limb evolves along all three axes as the various zones of activity work in concert. **D**, Under the guidance of the *wingless-type (Wnt)* signaling center, the limb differentiates in the dorsoventral axis. Ultimately, the digits separate under the influence of the *AER*, resulting in the familiar form of a human arm and hand.

is an inducer of AER-secreted FGF4 through a positive-feedback loop. Thus growth is exquisitely regulated in both the proximodistal and craniocaudal axes.

The dorsoventral axis is controlled in part by yet another set of genes located in the *Wnt* signaling center.^{1,3,4,6} The *Wnt7A* gene, expressed specifically in the dorsal ectoderm, is thought to play a significant role in dorsoventral polarity of the limb. The dorsal ectoderm where *Wnt7A* is expressed maintains the expression of *SHH*, thus integrating with the other two axes in the three-dimensional development of the upper limb.

TISSUE-SPECIFIC DEVELOPMENT OF THE UPPER LIMB

The upper extremity comprises multiple tissues, including bone, cartilage, muscle, tendon, vessels, nerves, and integument. Musculoskeletal development begins at the core of the limb bud. The chondrogenic core cells first differentiate into either chondrocytes or osteoblasts. Chondrification of the upper limb advances in a proximal to distal direction, beginning with the humerus and ending with the proximal parts of the distal phalanges. Important participants in this process include members of the sex determining region Y box (SOX) transcription factor group, the transforming growth factor beta (TGF-beta) superfamily, the FGF family, and the SHH protein. Joint development is the result of repression of chondrogenesis at specified joint sites.⁵ One of the proteins thought to be extremely important and upregulated at sites of joint development is Wnt14.¹² The muscles of the upper limb are identified in the embryo by week 7 of development. Paralleling skeletal development, proximal muscles evolve before more distal muscle groups, and superficial muscles develop before deeper muscles develop. Innervation of the developing muscles similarly begins at the limb bud and progresses distally. The vascular supply to the evolving limb begins as a series of capillary networks. The first vessel that appears is a marginal vein on the ulnar side of the developing limb. It is preserved as the subclavian-basilic system. The subclavian-brachial artery is the first artery to appear. It ultimately gives rise to the median artery, which initially supplies the hand. Although the ulnar artery appears before the radial artery, both finally lend vascular inflow to the hand as the median artery regresses.

MOLECULAR GENETICS OF CONGENITAL HAND ANOMALIES

Recent advances in molecular genetics have increased our understanding of the mechanisms involved with the organization and development of the upper extremity and of the genetic causes of some hand anomalies. On a molecular level, upper limb development occurs in a series of well-orchestrated biochemical steps. A cell produces and secretes a protein that binds a designated receptor on a target cell. The protein-receptor bond triggers changes within the target cell, resulting in specific transcription factors binding to DNA in the nucleus of the target cell. This leads to the expression of new target genes. When these sequences of events occur as planned, they culminate in the spatial development of the upper limb and hand.¹³

Several genes are involved in the normal development of the upper limb, and many are beginning to be identified. Mutations in many of the genes are associated with specified hand anomalies. Genes involved in preaxial polydactyly, split hand, and brachydactyly type C have been mapped to chromosomes, and the first human gene mutation associated with a hand anomaly is the *homeobox D13 (HOXD13)* gene mutation.^{1,14} As our understanding of hand development at the molecular level expands, more anomalies will be described genetically.

An introduction to the genetics of hand development and the anomalies that occur when the process diverges from the expected requires a cursory understanding of the homeobox genes.^{1,3,14} *HOX* genes are intimately involved in embryogenesis and are highly conserved across species. They direct cells within the early embryo into fields, each with the potential to become differ-

ent tissues and organs. All vertebrates have *HOX* genes organized into four complexes (*HOXA*, *HOXB*, *HOXC*, and *HOXD*), and each complex is located on a separate chromosome. The name *HOX* identifies a gene from one of the homeobox gene clusters. During upper limb development, the *HOX* genes play an important role in the interactions between the AER and the underlying PZ. As the limb evolves, various combinations of the *HOX* proteins are expressed at specific times and positions within the PZ. For example, the expression of the *HOXA* and *HOXD* is regulated in phases associated with the patterning of the segments of the upper limb: the arm (the embryologic name is *stylopod*), forearm (*zeugopod*), wrist (*mesopod*), and hand (*autopod*).^{5,7} In this way, the cellular milieu is directed proximodistally to evolve into the upper arm, lower arm, and hand structures. *HOXD* gene expression patterns in the hand result in five fields that lead to the morphologically distinct five recognized digits. For this reason, hand anomalies, such as syndactyly and polydactyly, may originate in the *HOXD* cluster.

Synpolydactyly is a congenital hand anomaly linked to a *HOXD* gene.¹⁴ Identified as syndactyly of the third and fourth fingers and the fourth and fifth toes in association with polydactyly of the same fingers and toes, synpolydactyly typically has an autosomal dominant inheritance pattern. The responsible gene was initially mapped to chromosome 2q31, and further work identified that the specific mutation was an expansion of a polyalanine sequence in the *HOXD13* gene, the first identified in humans. Similarly, the precise genetic causes of preaxial polydactyly, split-hand/split-foot malformation, and brachydactyly type C are beginning to be understood, each of which is mapped to chromosomes 7, 10, and 12, respectively. Current efforts are focused on identifying the specific mutations responsible for their occurrences.

As with all facets of medicine, advances in molecular genetics are just beginning to result in findings to complement our understanding of various anomalies and disease states. Our understanding of hand development and the anomalies that may occur will only be augmented as more data become available.

CLASSIFICATION SYSTEMS FOR CONGENITAL HAND ANOMALIES

Classification systems for pathologies and anomalies are essential. They provide a way for professionals to comprehensibly communicate with each other using consistent nomenclature to group similar pathologies together and to separate distinct pathologies, often indicating etiologic factors, management, and prognosis. Ideally, classification systems stratify conditions according to severity and the possible available treatments. In theory, they should guide users to an understanding of the nature of the disease process in question. However, because classifications are artificial and based only on the understanding of the pathology at a given point in time, they are inevitably subject to inaccuracies, changes in our knowledge, new understandings, and the variations of contemporary and future attitudes, only some of which will stand.

The history of the classification of congenital hand anomalies dates to the 1830s and the work of the French comparative anatomist Isidore Saint-Hilaire,¹⁵ who introduced names such as *phocomele*, literally meaning *seal hand*. After this publication, many other classification systems were developed. In 1974 Kelikian¹⁶ further described the timeline of congenital hand classification systems. The most commonly used and well-known classification system may be the one that Swanson and colleagues¹⁷ proposed in 1968 and that the International Federation of Societies for Surgery of the Hand subsequently revised and adopted.¹⁸ The classification contains seven categories (Box 41-1). The first two categories (failure of formation and failure of differentiation) contain the conceptual seeds for describing the causes of the nature of the malformations; however, the next five categories (duplication, overgrowth, undergrowth, constriction band syndromes, and generalized anomalies and syndromes) are essentially descriptive.

Failure of parts to form includes transverse and longitudinal deficiencies of the upper extremity. They are discussed in Chapter 42. Congenital transverse deficiencies are usually isolated,

Box 41-1 Swanson Classification of Congenital Hand Anomalies

- I. Failure of formation of parts
 - A. Transverse deficiencies
 - B. Longitudinal deficiencies
 - 1. Phocomelia
 - 2. Radial
 - 3. Central
 - 4. Ulnar
- II. Failure of differentiation
 - A. Synostosis
 - B. Radial head dislocation
 - C. Symphalangism
 - D. Syndactyly
 - E. Contracture
- III. Duplication
 - A. Thumb
 - B. Triphalangism
 - C. Polydactyly
 - D. Mirror hand
- IV. Overgrowth
 - A. Limb
 - B. Macrodactyly
- V. Undergrowth
- VI. Congenital constriction band syndrome
- VII. Generalized skeletal anomalies

sporadic, and unilateral and are identified according to the last remaining bone segment. The most common congenital transverse deficiency is a short below-elbow amputation. Longitudinal deficiencies may include radial, ulnar, or central deficiencies. Radial deficiencies affect the cranial, preaxial border of the arm and are often associated with systemic conditions, including Holt-Oram syndrome, thrombocytopenia-absent radius (TAR) syndrome, Fanconi anemia, and the VACTERL (vertebral defects, anal atresia, cardiac defects, tracheoesophageal fistula, renal anomalies, and limb abnormalities) association. The seriousness of the radial defect ranges from a minimally affected thumb to the absence of the thumb, carpus, and radius¹⁹ (Table 41-1). Ulnar deficiencies are far less common than their radial counterparts and affect the postaxial side of the limb. Although they may be associated with other musculoskeletal anomalies, they are not typically associated with systemic syndromes.

In 1993 Bayne²⁰ described the classification system that is probably most commonly used. Similar to the classification for radial dysplasia, it categorizes dysplasia according to the severity of the abnormality of the ulna (Box 41-2). According to findings at the thumb and first web space, ulnar deficiencies are further classified into four groups, ranging from a normal thumb and web space to an absent thumb²¹ (Table 41-2). Finally, central hand deficiencies, or a cleft hand, result from the longitudinal loss of the index, long, and/or ring rays of the hand.

Syndactyly is an abnormal connection between adjacent digits and is the most common congenital hand anomaly associated with failure of differentiation. The connection may involve the entire length of the digit (complete), or it may end proximal to the fingertip (incomplete) (Chapter 44). Furthermore, only skin and fibrous tissue may be involved (simple), or adjacent phalanges may be attached (complex). Congenital syndactyly occurs in 2 of every 10,000 live births,²² tends

Table 41-1 Classification of Radial Deficiency

Type	Thumb Anomaly	Carpal Anomaly	Distal Part of Radius	Proximal Part of Radius
N	Abs/hypo	Normal	Normal	Normal
0	Abs/hypo	Abs/hypo	Normal	Normal, radioulnar syndrome or radial head dislocation
1	Abs/hypo	Abs/hypo	>2 mm shorter than ulna	Normal, radioulnar syndrome or radial head dislocation
2	Abs/hypo	Abs/hypo	Hypoplasia	Hypoplasia
3	Abs/hypo	Abs/hypo	Absence of physis	Variable hypoplasia
4	Abs/hypo	Abs/hypo	Absence	Absence

Abs/hypo, Absent/hypoplastic.

Box 41-2 Classification of Ulnar Deficiency in the Forearm

1. Ulnar deviation of the wrist without hypoplasia of the ulna
2. Hypoplasia of the ulna
3. Partial absence of the ulna with radial bowing
4. Total absence of the ulna

Table 41-2 Classification of Ulnar Deficiency According to Abnormality of First Web Space

Type	Grade	Characteristics
A	Normal	Normal web space and thumb
B	Mild	Mild deficiency of first web space and mild thumb hypoplasia Intact opposition and extrinsic muscle function
C	Moderate-severe	Moderate-severe deficiency of first web space and thumb hypoplasia with malrotation, loss of opposition, and extrinsic dysfunction
D	Absent	Absence of thumb

to be familial, and may be isolated or part of more systemic syndromes, such as Apert syndactyly, which is discussed in Chapter 45.

Duplications include polydactyly, thumb duplications, triphalangism, and mirror hand are described in Chapter 46. Polydactyly occurs on both the preaxial and postaxial sides of the hand. Preaxial polydactyly (radial) is more common in whites, whereas postaxial polydactyly (ulnar) has a higher incidence in blacks. Postaxial polydactyly is often an autosomal dominant trait. In both preaxial and postaxial polydactyly, the extra digit may be a rudimentary element of soft tissue, or it may be nearly completely formed, including bones. Thumb duplications, more common in whites, are usually unilateral, sporadic, and isolated, and the classification is presented later in the chapter.

Macrodactyly, or overgrowth, is usually an isolated anomaly, but it is sometimes associated with other conditions such as neurofibromatosis or Klippel-Trenaunay syndrome. It is discussed in detail in Chapter 47.

Thumb hypoplasia is the most common form of undergrowth in developing hands (see Chapter 48). Blauth²³ has proposed a distinct and useful classification that is discussed in the section Classification of Specific Anomalies later in the chapter.

Congenital constriction band syndrome describes the demise of distal developing hand structures because of tourniquet constriction by amniotic bands in utero. Parts of the upper extremity, most often digits, are amputated. The level of amputation directs intervention and therapy, which may range from physical therapy to surgical intervention to optimize hand function. The classification, causes, and management are discussed in Chapter 49.

Evolution of the New Oberg, Manske, and Tonkin Classification System

With an increased understanding of the embryologic processes involved in upper extremity development, a renewed interest in classifying congenital anomalies arose. This is quite logical, because, at the time of inception of the Swanson classification,¹⁸ relatively little was known about the developmental biology of the patterning of the upper limb; thus it did not necessarily correlate well with the embryologic mechanisms leading to the disorders. Furthermore, the classification system provided no information on severity, prognosis, and management; it combined disorders that might intuitively be considered disparate; and it separated disorders that might otherwise be considered similar. However, it accomplished the difficult task of encompassing almost the entire, broad range of congenital hand anomalies. Because of its relatively simple nature, it was easy to use and apply practically. As a result, it was widely adopted as the classification of choice to describe congenital hand anomalies.

After the Swanson/IFSSH classification was generally accepted (see Box 41-1), understanding of the embryology underlying limb development and abnormal development evolved significantly, in part because the developing limb bud was the organ of choice for studying developmental patterning in general. In 1969 Wolpert²⁴ proposed the French Flag model of embryonic development, in which positional patterning information is determined by molecular gradients. Seminal work by Saunders,²⁵ Tickle,^{26,27} and others in the late sixties and later described the concept of the vertebrate limb patterned in three axes. This work was vital to our conceptual understanding of how the limb is patterned and therefore how congenital anomalies arise. Despite these dramatic scientific changes in our understanding and the development of other classification modifications,²⁸ the congenital hand surgery community continued to primarily use the clinical nomenclature of the Swanson/IFSSH classification for more than 3 decades.¹⁷

Recently, however, the developmental biologist Oberg, the late Paul Manske, and the Australian surgeon Tonkin (OMT)²⁹ reconsidered how we classify upper limb anomalies. They aimed to produce a comprehensive classification of upper limb anomalies that included our current understanding and nomenclature of the developmental biology of the growing limb bud³⁰ (Box 41-3). The OMT classification system²⁹ was proposed in 2010 and revised in 2013.³¹ Although it may at first seem very complicated, it is essentially divided into four major groups. The first group, the malformations, includes anomalies in formation and differentiation in which abnormal development has occurred along one or more of the three axes of development (for example, radial longitudinal deficiency). The second group, the deformations, describes an alteration or distortion to a limb or limb element that has already formed, such as those that occur with constriction bands. The third group includes dysplasias, in which “axial” and positional development is normal, but tissue growth is abnormal (hypertrophy, vascular anomalies, and other congenital tumorous conditions). The fourth group describes limb abnormalities associated with syndromes, such as Apert syndrome.

Box 41-3 Extended Version of the Oberg, Manske, and Tonkin Classification and Proposed Additions

- I. Malformations
 - A. Failure of axis formation/differentiation—entire upper limb
 1. Proximodistal axis
 - a. Brachymelia
 - i. Brachymelia with brachydactyly
 - ii. Symbrachydactyly
 - iii. Transverse deficiency
 - (a) Amelia
 - (b) Clavicle
 - (c) Humeral (long/short above elbow)
 - (d) Forearm (long/short below elbow)
 - (e) Wrist
 - (f) Metacarpal
 - (g) Proximal/distal phelans
 - (h) Phocomelia (total proximal/distal)
 - iv. Intersegmental deficiency
 2. Radioulnar (AP axis)
 - a. Radial longitudinal deficiency, thumb hypoplasia (with proximal limb involvement)
 - b. Ulnar longitudinal deficiency
 - c. Ulnar dimelia
 - d. Radioulnar synostosis
 - e. Humeroradial synostosis, elbow ankylosis
 3. Dorsoventral axis
 - a. Nail-patella, Furlong, and Al-Awadi syndromes
 - b. Arthrogryposis
 - c. Absent/ hypoplastic extensor-flexor muscles
 4. Unspecified axis
 - a. Undescended scapula (Sprengel)
 - b. Abnormal shoulder muscles
 - c. Complex abnormalities of the cervical spine and shoulder
 - B. Failure of axis formation/differentiation—hand plate
 1. Proximodistal axis
 - a. Brachydactyly
 - i. Metacarpal level
 - ii. Phalangeal level
 - iii. Brachysyndactyly
 - b. Symbrachydactyly
 - c. Transverse deficiency
 - i. Wrist
 - ii. Proximodistal carpal row
 - iii. Metacarpal
 - iv. Proximodistal phalanx
 2. Radioulnar axis (AP axis)
 - a. Radial (thumb) deficiency (no radius involvement)
 - b. Ulnar deficiency (no ulna involvement)
 - c. Radial polydactyly
 - d. Triphalangeal thumb
 - e. Ulnar polydactyly
 3. Dorsoventral axis
 - a. Dorsal dimelia (palmar nail)
 - b. Hypoplastic/aplastic nail
 - c. Arthrogryposis/windblown hand

Continued

Box 41-3 Extended Version of the Oberg, Manske, and Tonkin Classification and Proposed Additions—cont'd

4. Unspecified axis
 - a. Soft tissue
 - i. Syndactyly
 - ii. Camptodactyly
 - iii. Thumb-in-palm deformity
 - iv. Deviated finger without bony deformity
 - b. Skeletal deficiency
 - i. Clinodactyly
 - ii. Kirner deformity
 - iii. Metacarpal and carpal synostosis
 - iv. Carpal synostosis
 - v. Metacarpal synostosis
 - vi. Phalangeal synostosis
 - c. Complex: Syndactyly with synostosis of the phalanges
 - i. Cleft hand
 - ii. Synpolydactyly—central
 - iii. Apert hand
- II. Deformations
 - A. Constriction ring sequence
 - B. Trigger digits
 - C. Not otherwise specified
- III. Dysplasias
 - A. Hypertrophy
 1. Whole limb
 - a. Hemihypertrophy
 - b. Aberrant flexor/extensor/intrinsic muscles
 2. Partial limb
 - a. Macroductyly
 - b. Aberrant intrinsic muscles of the hand
 - B. Tumorous conditions
 1. Vascular
 - a. Hemangioma
 - b. Malformation
 2. Neurologic
 - a. Neurofibromatosis
 3. Connective tissue
 - a. Juvenile aponeurotic fibroma
 - b. Infantile digital fibroma
 4. Skeletal
 - a. Osteochondromatosis
 - b. Enchondromatosis
 - c. Fibrous dysplasia
 - d. Epiphyseal abnormalities
- IV. Syndromes
 - A. Specified
 1. Apert
 2. Arthrogryposis
 3. Baller-Gerold
 4. Bardet-Biedl

The IFSSH has reviewed the OMT classification system and recently decided to support its use instead of the previous system (the Swanson/IFSSH classification system).³² They claimed that the new system not only comprehensively covers the range of congenital malformations, but also accounts for the nature of the underlying developmental pathology in more detail than the previous classification system. Table 41-3 provides a comparison of these two classification systems. Recently, other studies have compared the new system with the previous one.^{30,31,33} The results supported the IFSSH findings—the OMT system comprehensively and, of most importance, dependably (that is, with reference to interobserver and intraobserver consistency) covers the range of congenital hand malformations. The authors cautioned that the OMT classification had not yet gained as widespread a popularity as its predecessor, perhaps because of the simplicity and entrenched popularity of the older classification. They explained that although the new classification system encompasses the spectrum of congenital upper limb conditions comprehensively, so does the Swanson/IFSSH system,³² but without the same degree of reference to the underpinning mechanisms. Neither classification system describes (or claims to describe) severity (or prognosis) or management. Both classifications present disorders that are not conveniently assigned to only one group or another; for example, Apert hand could be considered a failure of differentiation (Swanson classification), a malformation (OMT classification), and a syndromic hand.

How the OMT classification system stands the test of time, which its predecessor did remarkably well, will depend on how well it organically incorporates new disorders and further advances

Table 41-3 Comparison of the Swanson/IFSSH and Oberg, Manske, and Tonkin Classifications

Swanson/IFSSH Classification	Oberg, Manske, and Tonkin Classification
Type I: Failure of formation • Transverse • Longitudinal Type II: Failure of differentiation • Soft tissue • Skeletal • Tumors Type III: Duplication Type IV: Overgrowth Type V: Undergrowth Type VI: Constriction band syndromes Type VII: Generalized anomalies and syndromes	I. Malformations A. Abnormal axis formation/differentiation entire upper limb 1. Proximodistal axis 2. Radioulnar (AP axis) 3. Dorsoventral 4. Unspecified B. Abnormal axis formation/differentiation hand plate 1. Proximodistal axis 2. Radioulnar (AP axis) 3. Dorsoventral 4. Unspecified II. Deformation III. Dysplasias A. Hypertrophy 1. Whole limb 2. Partial limb B. Tumors 1. Vascular 2. Neurologic 3. Connective tissue 4. Skeletal IV. Syndromes A. Specified B. Unspecified

IFSSH, International Federation of Societies for Surgery of the Hand.

in developmental biology, and perhaps (of even more importance) how easily current and future pediatric upper limb surgeons will manage to apply it in daily practice.

Classification of Specific Anomalies

A review of the literature relating to almost any hand disorder (congenital, acquired, pediatric, and adult) will reveal multiple classification systems—too many to discuss in a single chapter. However, some systems are of special interest because of the relatively common nature of the conditions they describe and because of their longevity. These are briefly mentioned in the following sections.

Thumb Duplication

Wassel³⁴ described the most commonly used classification of thumb duplication while he worked as Adrian Flatt's fellow (1969). It describes the proximodistal skeletal level of the duplication (I through VI), and VII refers to a triphalangeal thumb³⁴ (Fig. 41-3). It is easy to use because of its simplicity and correlation with the necessary surgical management. However, because of the immaturity of the bones, the true skeletal nature of the duplication often cannot be determined based on radiographic and clinical assessments but only intraoperatively. The word *duplication* may be misleading in that both of the “duplicated” ray digits are invariably evidently smaller and more hypoplastic than a normal thumb (in contrast to ulnar duplication/polydactyly) and are perhaps more akin to a split or bifid thumb.

Ulnar Duplication and Polydactyly

In 1963 Stelling³⁵ and Duran et al³⁶ presented one of the most often used systems for classifying ulnar polydactyly (Table 41-4). It is easy to use. However, despite including most scenarios, it does not account for the nuances of many variations, such as the details of the relationship between the accessory and the main digit. As a result, a variety of other classification systems were proposed³⁶ (see Table 41-4).

Syndactyly

Temtamy and McKusick³⁷ published a classification for syndactyly that identified the affected web space (usually the third). Swanson¹⁸ presented a descriptive classification of syndactyly based on

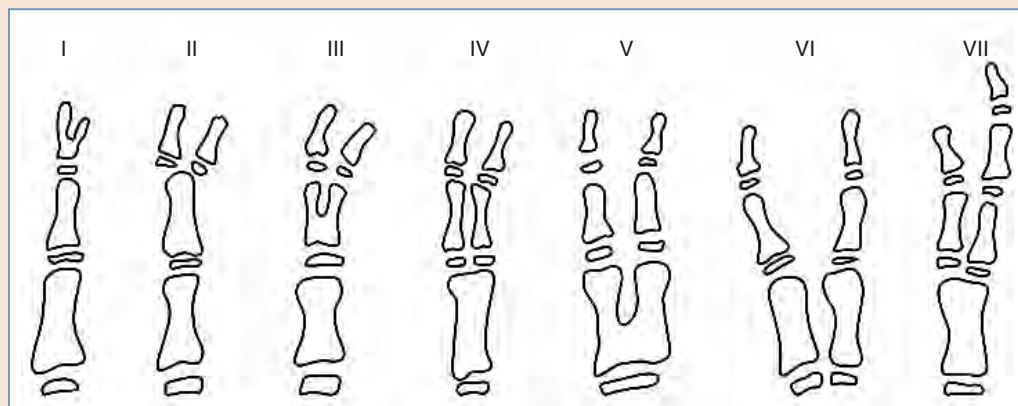


Fig. 41-3 The Wassel classification of thumb duplication.

severity (simple, with skin alone, and complicated, with the involvement of other tissues). Variations were published. Currently, the most commonly used adaptation of this classification for syndactyly describes the pathology as complete (in a proximodistal direction), incomplete, simple (not involving bone), complex (involving bone), or complicated (an abnormal arrangement of structures beyond fusion).^{38,39}

Table 41-4 Previously Reported Classification Systems for Ulnar Polydactyly and the Proposed Classification System in the Current Study

Temtamy-McKusick Classification*	Stelling Classification†	Rayan-Frey Classification‡	Al-Qattan Modification of Rayan-Frey Classification§	Pritsch et al Classification for Type A Ulnar Polydactyly¶	Duran et al Current Classification
Type A: A well-formed extra digit with an articulation	Type I: Soft tissue only	Type I: A soft tissue nubbin	Type I: A small, soft tissue nubbin with no bone or nail	Type I: A fully developed sixth ray that articulates separately with carpals	Type I (simple type): A skin nub similar to a verruca that does not contain bone and nail or a nonfunctional digit that contains bone or nail, or both, and a small pedicle that does not articulate with the metacarpal
Type B: A poorly formed extra digit connected to the hand by a skin bridge	Type II: Duplication of phalanges	Type II: A pedunculated, nonfunctioning digit	Type II: A pedunculated, nonfunctioning digit attached to the hand by a narrow (<3 mm) pedicle (type IIA) or by a wider pedicle (type IIB)	Type II: An extra digit on the lateral side of the fifth digit with an intercalated distal metacarpal remnant	Type II (hypoplastic type): a hypoplastic proximal phalanx
	Type III: Complete duplication of the phalanges and metacarpal	Type III: A well-formed, functioning digit articulating with a bifid fifth metacarpal head or fused to the fifth metacarpal at a right angle or hypoplastic absence of the proximal phalanx	Type IIIA: A well-formed, functioning digit articulating with a bifid metacarpal head or a partially duplicated fifth metacarpal Type IIIB: A well-formed, functioning digit with its proximal phalanx fused to the fifth metacarpal (type IIIB)	Type III: A supernumerary digit arising from a hypoplastic sixth metacarpal or fused to a fifth metacarpal	Type III (malformations at the proximal phalanx level, two subdivisions): Type IIIA—a bifid proximal phalanx Type IIIB—a duplicated proximal phalanx

*Does not include fusion between the proximal phalanx and metacarpal, triplication, polysyndactyly, or hypoplastic proximal phalanx.

†Does not include fusion between the proximal phalanx and metacarpal; a bifid metacarpal head, metacarpal remnant, or bifid metacarpal shaft; triplication; or polysyndactyly.

‡Does not include duplications of the proximal phalanx or triplication of the little finger.

§Does not include duplications of the proximal phalanx.

¶Does not include triplication or polysyndactyly.

Continued

Table 41-4 Previously Reported Classification Systems for Ulnar Polydactyly and the Proposed Classification System in the Current Study—cont'd

Temtamy-McKusick Classification*	Stelling Classification†	Rayan-Frey Classification‡	Al-Qattan Modification of Rayan-Frey Classification§	Pritsch et al Classification for Type A Ulnar Polydactyly¶	Duran et al Current Classification
		Type IV: Complete duplication with a separate sixth metacarpal	Type IV: Duplication with a separate sixth metacarpal	Type IV: An extra digit originating from the fifth metacarpophalangeal joints	Type IV (malformations at the metacarpal level): Type IV A—fusion between the proximal phalanx and metacarpal Type IV B—a bifid metacarpal head Type IV C—a metacarpal remnant Type IV D—a bifid metacarpal shaft or a complete metacarpal duplication
		Type V: Polysyndactyly	Type V: Other cases, including polysyndactyly and triplication of little finger	Type V: Extra digit originates from bifid fifth proximal phalanx	Type V (complicated type): Described as triplication of little finger or polysyndactyly or coexistence of both

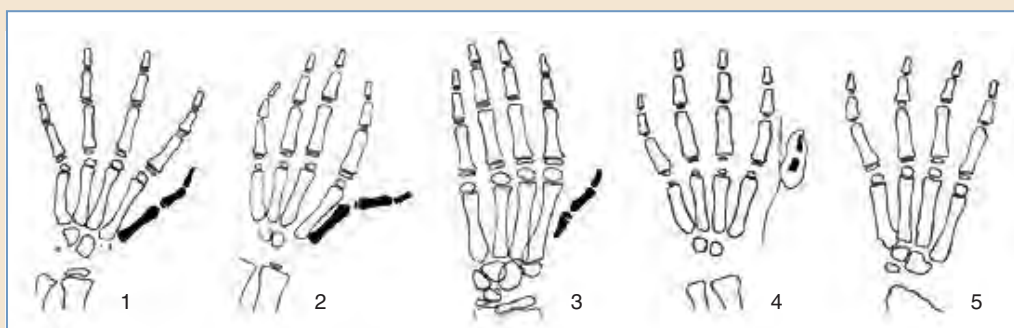
*Does not include fusion between the proximal phalanx and metacarpal, triplication, polysyndactyly, or hypoplastic proximal phalanx.

†Does not include fusion between the proximal phalanx and metacarpal; a bifid metacarpal head, metacarpal remnant, or bifid metacarpal shaft; triplication; or polysyndactyly.

‡Does not include duplications of the proximal phalanx or triplication of the little finger.

§Does not include duplications of the proximal phalanx.

¶Does not include triplication or polysyndactyly.

**Fig. 41-4** Classification of congenital thumb hypoplasia.

Thumb Hypoplasia

Blauth²³ described the classic classification system, in which thumb hypoplasia is categorized according to severity. Various modifications have been developed⁴⁰ (Fig. 41-4). Manske and McCarroll⁴¹ introduced a division in type 3, which may be the key modification. Type 3A is a thumb with a functioning carpometacarpal joint, and type 3B does not have a functioning carpo-

metacarpal joint. The significance of this division is that types 1 to 3A describe thumbs that can probably be reconstructed to a useful digit, whereas types 3B to 5 cannot. David Chiu⁴² recently presented the possibility of reconstructing type 3B and those even more severe using toe-to-hand transfer and carpometacarpal joint reconstruction using a tendon coil. Long-term outcomes have not been reported.

CONCLUSION

The spectrum of congenital hand anomalies is interesting and varied. Our understanding of the causes of these conditions continues to grow. Surgically, the challenges of creating an organ as intricately engineered as the hand is daunting but nonetheless rewarding as we strive to deliver a functional hand to an infant born with one of these anomalies. Central to this endeavour is an understanding of hand embryology. This is needed to optimize strategies for therapy and intervention and to classify this diverse group to facilitate communication between health care professionals working with children with congenital upper limb anomalies.

KEY POINTS

- The upper limb develops from day 26 (limb buds emerge) to day 53 (five digits are formed) of gestation.
- Homeobox gene expression determines the craniocaudal position of limb development.
- Limb buds develop and differentiate in three axes: proximodistal, dorsopalmar, and radioulnar. Perturbations in this development can lead to congenital anomalies.
- The IFSSH currently recommends the OMT classification,²⁹ which is based on the developmental anomaly responsible for the abnormality (malformations, deformations, dysplasias, and syndromic). The previous IFSSH classification (proposed by Swanson and colleagues¹⁷ and still commonly used) is primarily based on the morphology of the anomaly.
- Both systems should be appreciated, including their evolution.
- Specific congenital anomalies have their own classifications. Many are graded according to severity and provide indications for treatment options.

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42

Congenital Hand Anomalies: Failure of Formation

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he group of conditions classified based on the failure of formation of parts is characterized by complete or partial arrest in development of a limb.¹ These conditions are further classified as those with transverse arrest and those with longitudinal arrest. Transverse arrest refers to conditions with terminal transverse deficiency usually presenting with a stump at the level of amputation (Fig. 42-1). All other skeletal limb deficiencies are longitudinal (Fig. 42-2).

In several studies evaluating the frequency of failure of formation of the upper extremity, estimates range from 10.9% to 17.6%.²⁻⁵ Giele et al² conducted a survey in Western Australia of data over an 11-year span and concluded that failure of formation of parts accounted for 15% of all congenital upper extremity anomalies. A similar study in Sweden documented that failure of formation occurred in 103 of 585 congenital upper limb anomalies (17.6%) in children born between 1997 and 2007.⁵

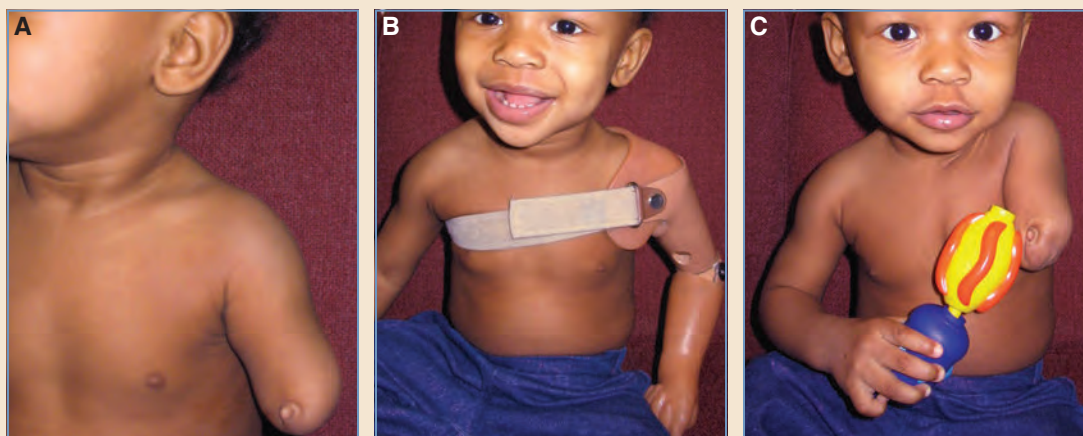


Fig. 42-1 **A**, This 13-month-old child has transverse arrest at the level of the distal humerus. The nubbin on the anterior surface of the distal aspect of the limb may represent the remaining, undeveloped limb. **B**, The child is wearing a passive prosthesis on her left arm. The primary purpose of this prosthesis is to normalize the appearance of the limb. However, the device prohibits the child's sensation of the limb. **C**, With the prosthetic device removed, the child integrates her left arm for bilateral play. She uses the available range of motion (ROM) and sensory feedback on the affected side to assist the unaffected limb.



Fig. 42-2 This radiograph shows the upper extremity of an infant with unilateral absence of the ulna. This is classified as postaxial longitudinal arrest (or ulnar deficiency).

FUNCTIONAL EVALUATION

Functional evaluation of children with congenital hand anomalies not only involves a detailed musculoskeletal examination, but also a thorough evaluation of how they use their upper extremities in daily activities.⁶ A thorough functional evaluation is critical in making recommendations for nonsurgical and surgical treatment.

At the initial visit, the medical and family history should be documented. Concern about inheritance may warrant a referral to a genetics professional. A thorough musculoskeletal examination of both upper extremities is conducted, noting in detail the present and absent skeletal structures and active and passive ROM. The lower extremities are evaluated for associated anomalies. Caregivers are informed of the etiologic factors and prognosis of their child's condition. The family's general concerns regarding cosmesis and overall psychosocial response to the child's condition are noted.

Regular evaluation begins when children are 3 months of age and includes assessment of fine motor, bimanual skills and upper limb use in daily activities. This provides valuable information

for determining surgical management. Global development is evaluated during infancy to help caregivers have appropriate expectations for their child's development. Caregivers need reassurance of their child's ability to adapt and successfully achieve developmental milestones, despite the limb difference.

Children are evaluated when they are of preschool and school age to determine changes in ROM and performance in activities of daily living. Deficits in upper limb use or prehension in daily living, school, and/or leisure activities may provide indications for surgical or therapeutic reconstruction. Therapists can also provide input regarding adaptive techniques and aids to assist with daily activities. This is especially useful for patients who require assistance in adapting to a specific activity or sport. Routine evaluation is important, because functional demands change as patients mature.

Transverse Arrest

Transverse arrest is the congenital terminal amputation of a limb. It is classified according to the anatomic level where the limb terminates.^{1,7,8} The common levels of transverse arrest are the following⁶⁻⁸:

- Phalangeal
- Transmetacarpal
- Wrist
- Proximal forearm
- Midarm
- Shoulder

Amputation at the proximal forearm is most common.^{7,9,10}

Transverse arrest is usually unilateral^{7,11} and occurs more frequently on the left side.^{3,5,10} It is not usually associated with other malformations.¹¹ However, it has been confused with amniotic band syndrome.¹² The literature is varied regarding the incidence in males and females. Ogino et al³ reported no difference in incidence between gender. Birch-Jensen reported¹⁰ a female/male ratio of 3:2.

Epidemiology

Lamb et al¹³ conducted a survey of seven clinics internationally through the congenital malformations committee of the International Federation for Societies of the Surgery of the Hand. The extrapolated frequency of transverse arrest from these data was 1.5 in 100,000 live births. Birch-Jensen¹⁰ reported an incidence of 1 in 20,000 newborns for forearm arrest and 1 in 270,000 newborns for all transverse arrests. In another study, the incidence of transverse arrest was reported as 0.19 in 1000,¹⁴ accounting for 2.9% to 6.8%^{3-5,13} of children with congenital upper limb malformations.

A universal cause of congenital transverse arrest has not been identified.¹¹ Most cases are considered sporadic occurrences; however, the possible influence of teratogenic factors has not been ruled out.¹⁵ A positive family history has not been found.^{3,4} According to some authors, the mechanism of injury is linked to vascular injury in utero.^{16,17} Graham et al¹⁶ found that mechanical compression of the embryo at an early stage of development caused vascular disruption and ischemia, thereby arresting the development of the limb. Hoyme et al¹⁷ found evidence of vascular injury leading to ischemia in the developing limb. They reported that embolization from thrombi originating from the placenta caused occlusion in the brachial artery of the fetus.

Anatomy

Forearm amputation occurs most often in the proximal third of the forearm.¹⁰ The olecranon and trochlea are usually well developed; however, the development of the radial head varies.⁷ Subluxation of the proximal radioulnar joint and radial head dislocation are common.¹¹ The stump may have a variety of contours but is usually bulbous and well padded.^{8,9} Occasionally, vestigial nubbins with or without nail remnants are present at the end of the stump⁷⁻⁹ (see Fig. 42-1).

Patients with transverse amputation at the metacarpal or wrist levels may have motion at the radiocarpal joint if their extrinsic flexors and extensors of the forearm are well developed.¹¹ The remaining distal portion of the limb is often hypoplastic. Dimpling or retraction of tissue at the stump may indicate the presence of extrinsic flexor and extensor tendons; however, these structures are often hypoplastic.¹¹ Vestigial nubbins with or without nail remnants may be present.^{11,18}

Phalangeal amputations are rare⁷ and often associated with amniotic band syndrome^{11,12} (Fig. 42-3). If amniotic band syndrome is suspected, a thorough evaluation of all limbs should be conducted for evidence of constriction rings. Generally, all structures proximal to the level of the transverse arrest are normal, but osseous hypoplasia in the hand and forearm may occur.^{11,18}

Children with unilateral transverse arrest spontaneously and actively use their limb to explore and feel objects, because sensation is intact in the stump. Prehension is typically absent in the affected limb. However, many children use the limb to assist their dominant, unaffected limb for bilateral activities. For example, the affected limb is used to stabilize objects against the chest for manipulation with the unaffected hand. Some clinicians think that a passive upper extremity prosthesis (see Fig. 42-3) can assist in balance and gross motor development in infancy.¹⁹ Equalizing the limb length helps patients perform developmental skills¹⁹ such as creeping, propping in a prone position, and transitional movements from sitting to standing. We have observed these children achieve developmental milestones within an age-expected timeline without a passive prosthesis. Furthermore, families report that their children become frustrated with the passive prosthesis. We think this is at least in part from the inhibition of sensory exploration with the affected limb.

Evaluation

A child with transverse arrest should initially be seen within the first 3 months of birth for a functional assessment. Some parents are very keen to have vestigial nubbins removed to reduce the risk of accidental injury to these parts. We prefer to allow children to be involved in this decision at a later age. Some children seem to develop a fondness for their nubbins, whereas others find



Fig. 42-3 This 5-month-old infant has amniotic band syndrome affecting the digits in her right hand. Constriction bands are present around the proximal phalanx of her long and ring fingers. The distal and middle phalanges of the ring and little fingers are amputated. Digital amputation with evidence of constriction bands, as shown, is an indication of amniotic band syndrome as opposed to transverse arrest.

them inconvenient. Referral to an appropriate prosthetic facility can be initiated at this time. Although we have expressed some reluctance about early prosthesis use, referral to such facilities will provide parents with information about their child's present and future prosthetic options.

Nonoperative Management

Patients who have transverse arrest proximal to the forearm may be candidates for a prosthetic limb. Prosthetics are effective in providing a degree of prehension in the affected limb. However, young children may neglect their prosthesis because of the lack of sensory feedback and favor the sensate hand.²⁰ Visual cues and increased attentiveness are required of children who use a prosthesis.²¹

Shaperman et al²² surveyed data from 45 prosthesis clinics in North America regarding their clinical practice. The timing of the initial fitting of prosthetic devices was guided by the evaluation of the child's developmental signs such as evidence of independent sitting and exploration of the hand. The age at the initial fitting ranged from 2 months to 2 years. All clinics first introduced the children to passive prosthetic devices and then introduced an active device at an average age of 18 months (see Fig. 42-3). They found that a child's incorporation of a prosthetic device was not related to early fitting. Children with myoelectric prostheses participated in therapy programs to learn how to activate their terminal device.²³ As they matured, therapists assisted them and their family in using their myoelectric limb for specific activities in their daily living.²³ Children with short residual limbs often were unhappy with the distal weight of the prosthesis.²³

Amputations distal to the wrist may be fitted with a passive prosthetic hand or spatula (Fig. 42-4). Prosthetic hands are not functional devices, but they provide aesthetic benefit. A cosmetic hand is typically prescribed at adolescence for social functions.²¹ Spatula prosthetic devices are designed to allow children to oppose their hand against a hard surface for grasp.²⁴ These patients are not candidates for electronically powered prosthetic devices, because the amputation is too distal to support the mechanics of a myoelectric device.



Fig. 42-4 On the *left* is a spatula orthotic that can be used to facilitate prehension for a child with only one plane of movement. The spatula orthotic is secured to the child's forearm with Velcro straps. The child can press down on the surface of the spatula with their digits to hold small objects. In the *center* is an example of a silicone passive cosmetic prosthesis. On the *right* is a baseball glove fabricated to fit on a child's hypoplastic hand to facilitate participation in an activity of interest.

Operative Management

Operative options for managing transverse arrest are limited. We recommend removal of vestigial nubbins that interfere with normal activities. Ordinarily, this would be done once the child has expressed an interest in proceeding. However, an earlier procedure may be indicated if the nubbin causes a functional problem, such as recurrent bleeding with trauma.

The Krukenberg procedure is usually considered a reasonable option only in bilateral amputees (either congenital or acquired) who are also blind.²⁵ However, other authors have attempted to expand the indications for the procedure to include patients with intact sight, stating that the improvement in function outweighs the cosmetic trade-offs.^{26,27}

The procedure entails constructing a pincer grasp between the radius and ulna.²⁷ The pronator teres becomes the primary adductor of the radius. A cleft is created between the radius and ulna with dorsal and volar incisions, preserving full-thickness tissue on the tactile surfaces of the reconstruction. The flexor digitorum profundus and flexor pollicis longus muscle bellies are excised to debulk the stumps and to facilitate closure. The median and ulnar nerves are divided distal to the branches to the flexor digitorum superficialis and flexor carpi ulnaris. The flexor digitorum superficialis tendons, which help to motor the pincer grasp, are then evenly distributed between the radius and ulna and inserted into the bone. The tendons of the extensor digitorum communis are similarly distributed between the radius and the ulna, and the deep extensors are excised to facilitate closure. The interosseous membrane is then divided, opening the cleft between the radius and ulna as widely as possible. Split-thickness skin grafts are used as needed; only full-thickness skin should be used on the tactile surfaces.²⁶

Longitudinal Deficiency

Conditions classified as failure of formation with longitudinal deficiency are typically identified by the bones that are completely or partially absent.¹ They are classified as preaxial, central, postaxial, and intersegmental deficiencies.¹

The prevalence of longitudinal deficiencies is 4.6 in 10,000.¹³ Lamb et al¹³ and Ekblom et al⁵ reported an incidence of 12.5% and 13.8%, respectively, of total congenital upper limb malformations. Other studies have indicated lower incidences of 4.3% to 8%.^{3,4}

RADIAL RAY DEFICIENCY: PREAXIAL ANOMALY

Radial ray deficiency, also known as *radial club hand*, *radial hemimelia*, *radial dysplasia*, *radial aplasia*, *radial hypoplasia*, and *radial ray hypoplasia*,^{9,27} is characterized by shortening of the forearm and marked radial deviation of the hand at the wrist²⁸ (Fig. 42-5). The lack of radial support causes the hand and wrist to deviate toward the side of the deficiency.¹¹ The range of radial deficiency spans from minor abnormality of the thenar muscles to complete absence of all preaxial structures involving the upper arm and brachial plexus.¹¹

Riordan^{28a} classified radial ray deficiency into two main types of deformity: complete absence and partial absence of the radius. Bayne and Klug²⁹ have further classified this condition into four types²⁹ (Fig. 42-6 and Table 42-1). Type IV, complete absence of the radius, is most common. James et al³⁰ have described type 0, in which the radial carpus is hypoplastic and the wrist is deviated radially, but the radius itself is of normal length.

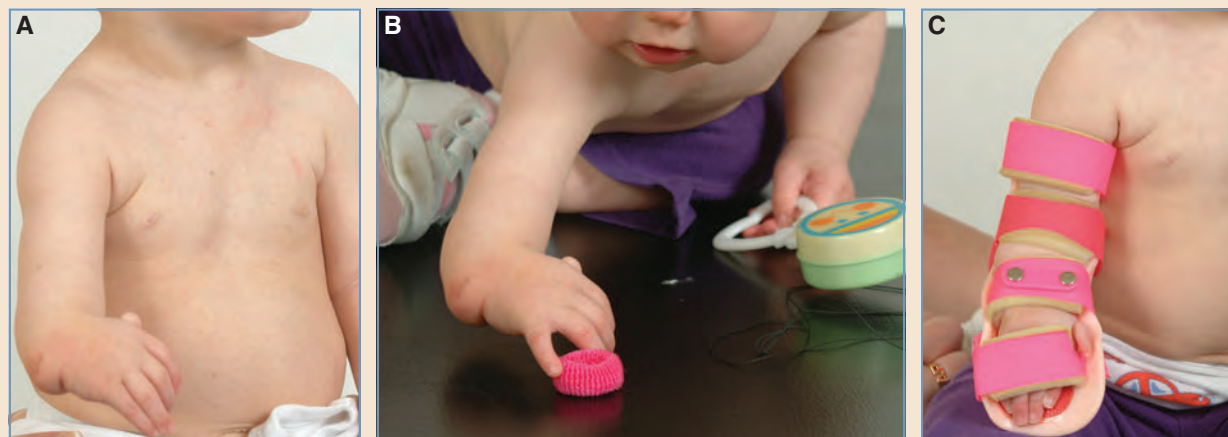


Fig. 42-5 A, This 2-year-old child has a typical presentation of a radial longitudinal deficiency type IV (see Table 42-1). She has marked radial deviation of the wrist on the forearm. (The elbow is held in extension in this image.) The residual ulna is shortened to approximately 60% on the unaffected side. This child also has a Blauth type IV hypoplastic thumb, otherwise known as a “pouce flottant” or floating thumb (see Chapter 43). B, As the child reaches for a small object, she widens the web space between her long and little fingers to prepare for grasp. The ulnar digits are preferred for prehension. C, The child is wearing a static low-temperature thermoplastic orthotic that centralizes the wrist over the ulna. For patients who may undergo centralization surgery, static splints can be used in conjunction with daily traction stretches to optimize passive ROM.

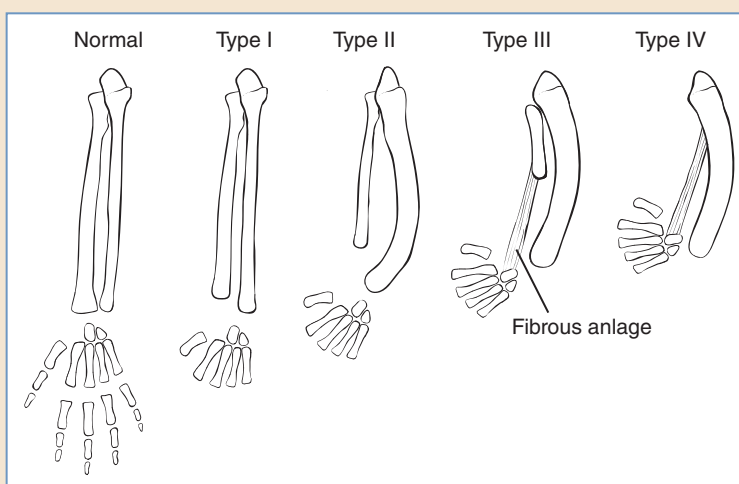


Fig. 42-6 The Bayne and Klug²⁹ classification of radial ray deficiency. The thumb may be hypoplastic in each type. Type I patients have a slightly short radius without significant angulation of the wrist and no bowing of the ulna. Type II patients have a hypoplastic radius, the ulna may be slightly bowed, and the hand is radially deviated at the wrist. In type III (the most common presentation), the wrist is tethered to the remnant of the radius by a fibrous anlage, resulting in progressive radial deviation of the wrist with growth. In type IV, the radius is absent, the ulna is bowed (although often less so than in type III), the hand is radially deviated at the wrist, and the elbow may be unstable.

Table 42-1 Bayne and Klug's Classification of Radial Ray Deficiency

Type	Characteristics
I Short distal radius	Decreased growth of the distal radial epiphysis causes a short radius.
II Hypoplastic radius	The proximal and distal epiphysis of the radius are hypoplastic.
III Partial absence of the radius	The defect can be in the proximal, middle, or distal third of the radius. The hand is radially displaced. The ulna is thickened, shortened, and bowed radially. The wrist is unsupported.
IV Total absence	The hand is unsupported and severely radially displaced.

Bilateral cases of radial ray deficiency make up 50%^{5,28a} or more²⁹ of all cases. Even patients considered to have unilateral deficiency on the basis of radial hypoplasia alone have some hypoplasia in the opposite extremity (for example, a hypoplastic thumb).²⁹ Furthermore, a higher incidence in the right limb has been reported in unilateral cases.^{28a,31} Some studies of radial ray deficiency have indicated a greater incidence in males than in females.^{29,31}

Epidemiology

Birch-Jensen¹⁰ reported an incidence of 1 in 30,000 births, and Platt³² reported 1 in 100,000 births. More recently, Ekblom et al⁵ and McGuirk et al¹⁴ reported an incidence of 0.13 and 0.14 in 1000 live births, respectively. Most cases have no positive family history.^{3,33} However, a few familial cases have been published.³³ Temtamy and McKusick¹⁵ indicated that most inherited cases are transmitted in an autosomal dominant pattern with marked variability in expression. Autosomal regressive inheritance has been suggested. The possibility of teratogenic¹⁵ and environmental³² influence in sporadic cases has not been ruled out. The general consensus is that no clear etiology, genetic, or environmental factors have been found.¹¹

The diversity of associated anomalies with radial ray deficiency have been well documented.^{8,15,33-35} Tsuyuguchi et al³⁵ found an associated anomaly in 46% of 26 patients with radial ray deficiency. More recently, Ekblom et al⁵ noted an associated anomaly in 60.6% of 33 children with radial ray deficiency. In addition, associated anomalies are often associated with bilateral cases. Upton¹¹ reported associated anomalies were present in 40% of unilateral cases and in 77% of bilateral cases. Temtamy and McKusick¹⁵ classified associated anomalies into 10 subcategories (Box 42-1). The most commonly reported associated syndromes are Fanconi pancytopenia syndrome, thrombocytopenia-absent radius syndrome, Holt-Oram syndrome, associations with craniofacial syndromes, and VACTERL (vertebral defects, anal atresia, cardiac defects, tracheoesophageal fistula, renal anomalies, and limb abnormalities).³⁴

Anatomy

The pattern of deficiency in the radial structures ranges from mild to severe involvement of the preaxial structures of the upper limb¹¹ (Table 42-2). James et al³¹ evaluated the pattern of deficiency in 124 patients with radial ray deficiency. In this study, distal deficiency often occurred in isolation. However, proximal deficiency was always associated with distal deficiency. Expression of deficiency generally moves from distal to proximal structures along the preaxial side of the limb. An exception to this rule was children with thrombocytopenia-absent radius syndrome. These children may present with an absent radius and a relatively well-developed thumb.^{31,32}

Box 42-1 Ten Subcategories of Associated Conditions With Radial Ray Deficiency

1. Radial defects with oral facial malformations
2. Radial defects with blood dyscrasias
3. Craniosynostosis–radial dysplasia syndrome
4. Radial defects with congenital heart disease: Holt-Oram syndrome
5. Radial defects with Duane anomaly
6. VACTERL
7. Radial defects with imperforate anus
8. Keutel-Kindermann syndrome
9. Chromosomal aberrations associated with radial defects
10. Thalidomide syndrome

Table 42-2 Commonly Absent and Abnormal Anatomy in Radial Ray Deficiency

Structure	Commonly Absent	Abnormal
Bone	Radius Scaphoid Trapezium Thumb metacarpal and phalanges	Scapula Clavicle Humerus Ulna Carpal bones
Muscle	Pectoralis major (clavicle or costal parts may be absent) Biceps (long head) Brachioradialis Anconeus Supinator Pronator teres Pronator quadratus Palmaris longus ECRL/ECRB EI Intrinsic/extrinsic muscles of the thumb	Pectoralis major Coracobrachialis Deltoid Brachialis Biceps (short head) Brachioradialis Supinator Pronator teres Palmaris longus ECRL/ECRB FCU/FCR FDM FDS/FDP EI EDC Intrinsic/extrinsic muscles of the thumb
Nerve	Myocutaneous nerve Radial nerve (distal to elbow)	Myocutaneous nerve Radial nerve Median nerve (innervates the radial aspect of the hand)

This table is by no means exhaustive, and many variations are known to occur.^{28a,29,33,36}

ECRL/ECRB, Extensor carpi radialis longus/extensor carpi radialis brevis; *EDC*, extensor digitorum communis; *EI*, extensor indicis; *FCU/FCR*, flexor carpi ulnaris/flexor carpi radialis; *FDM*, flexor digiti minimi; *FDS/FDP*, flexor digitorum superficialis/flexor digitorum profundus.

Proximally, the shoulder girdle can be affected in severe cases of radial ray deficiency.^{28a} The scapula may be reduced in size, and the clavicle is shorter and slightly more curved than normal. The humerus may be shortened and hypoplastic. Skeletal changes in the shoulder girdle may correspond with abnormal insertions of the pectoral major, pectoral minor, and deltoid muscles.

The long head of the biceps is frequently absent or abnormal.^{28a} When the long head of the biceps is present in patients with radial ray deficiency, it usually inserts into the lacertus fibrosus.³⁶ The short head of the biceps is always present but is usually fused with another muscle, such as the coracobrachialis or the forearm muscles.³⁶ The brachialis is usually present but may be normal, rudimentary, or fused.^{6,28a} The brachioradialis is usually absent if the radius is totally absent or is rudimentary with an abnormal insertion.^{28a,36} The joint ROM in the elbow may be impaired. Elbow stiffness in infancy may improve with time.³³ However, the absence of active elbow flexion is an important consideration for surgical management.^{29,33} Stiffness in elbow extension, along with absent or minimal active elbow flexion, is a contraindication for centralization or radialization of the wrist in patients with radial ray deficiency. Such procedures are not indicated, because the hand can reach midline at the face and body more easily in a radial position for daily activities.³⁷ Centering the hand on the forearm would worsen function.

In the forearm, if the radius is not totally absent, the proximal fragment of the bone is preserved. It can vary in size and shape and may or not be fused with the ulna.³⁶ When the radius is absent, the forearm typically achieves 60% of optimal growth.⁹ This proportion will remain throughout the child's development.⁹ The ulna is never normal in severe cases of radial ray deficiency.¹¹ This becomes more evident with severity of deformity as neurovascular and skeletal deficiencies become more pronounced.¹¹ The ulna is usually shortened, thickened, and curved, with the concavity toward the radial side^{28a,36} (Fig. 42-7). Muscle deficiencies in the forearm vary proportionately to the skeletal deficits.⁹ Muscles originating from the medial epicondyle (that is, the pronator teres and flexor carpi radialis) are usually well differentiated. However, they may have abnormal insertions, causing them to act as radial deviators of the hand.⁹ Alternatively, muscles originating from the lateral epicondyle are often affected. The brachioradialis, the extensor carpi radialis longus, and the extensor carpi radialis brevis are usually absent or abnormal. The supinator is typically absent, except in some cases in which the proximal radius is present.^{28a,36} The pronator teres may be absent if the radius is totally absent, or it may be rudimentary or fused with the biceps-brachialis mass, palmaris longus, or flexor carpi radialis when the radius is present.^{28a,36} The pronator quadratus is usually totally absent.³⁶

The length of the forearm has functional and cosmetic consequences. Patients are required to tailor their sleeves on clothing to accommodate shortened arm length (Fig. 42-8). Lamb³³ reported that their ability to be independent in toileting depends on their limb length, and that this especially affects patients who have bilateral involvement. Goldfarb et al³⁸ studied 21 adult patients (25 wrists) with radial ray deficiency, Bayne classification III or IV, who underwent centralization as a child (age average 22 months). The results suggested that the functional ability of these patients were more greatly affected by hand function than by wrist angulation or limb length. They indicated that many patients were able to compensate by using exaggerated trunk flexion to increase limb length. Of those treated in this study, seven patients were affected bilaterally, and four of these seven had bilateral centralization. Centralization of the wrist and hand on the ulna naturally provides some augmentation of limb length.³⁹

Radial deviation of the wrist on the forearm varies from 30 to 90 degrees.²⁸ The amount of radial deficiency is related to the degree of radial deviation, hand instability, and soft tissue deficiency.^{9,32,36} Patients with total absence of the radius have the most severe deviation and least function.^{28a} The radial carpal bones are frequently absent.^{28a,36} The scaphoid and trapezium are most often affected and are usually absent or fused to other bones.^{28a,36} With skeletal maturity, intercarpal fusions become more apparent.³³ Carpal deficiencies appear to be directly related to



Fig. 42-7 This radiograph shows the right upper extremity of a child with radial longitudinal deficiency type IV. The radius is absent, and the residual ulna is shortened and bowed.

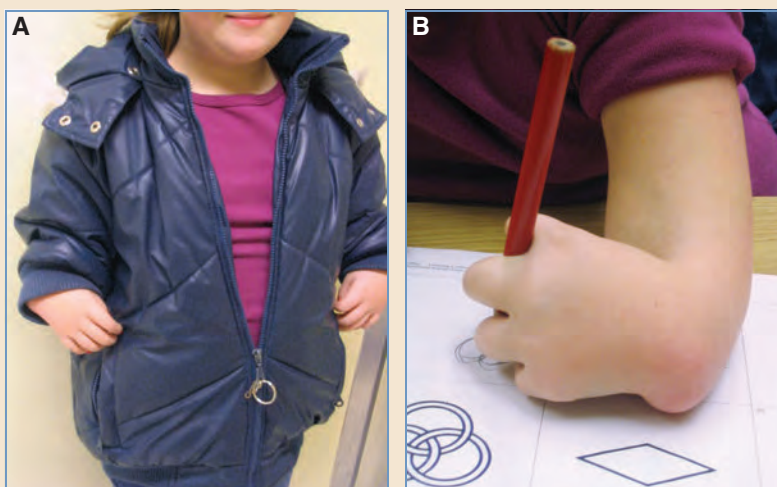


Fig. 42-8 **A**, The length of the sleeves of this winter jacket was custom tailored for this 10-year-old child with bilateral radial longitudinal deficiency type IV. An adaptive zipper ring was attached to the zipper pull to compensate for decreased strength and precision in fine grasp. **B**, The child demonstrates an adaptive pencil grasp. The pencil is held in an interdigital grasp with stabilization against the thumb.

the deficiency of the thumb.^{9,35} Isolated thumb hypoplasia is the distalmost expression of radial ray deficiency. The clinical presentation and management of isolated thumb hypoplasia is discussed in Chapter 43. Hand anomalies in radial ray deficiency are often confined to the thumb, index finger, and long finger.⁹ Patients with radial ray deficiency may also have a degree of hypoplasia or aplasia of the thenar muscles.^{9,35}

Classifications for isolated thumb hypoplasia, such as that presented by Blauth, can generally be used to describe the range of deficiencies in the thumb associated with radial ray deficiency (see Chapter 43). The first metacarpal and intrinsic muscles of the thumb are abnormal or absent, depending on the severity of thumb hypoplasia. Camptodactyly of the digits, especially the index and long fingers, often occurs with restriction in metacarpophalangeal joint flexion.⁹ The

pattern of digit deficiency progresses in severity toward the radial side of the hand. In more severe cases, multiple joints can be stiff, thereby resembling severe arthrogryposis.³³ The extrinsic finger flexors and extensors may be abnormal. The extensor indicis may be absent or abnormal.³⁶

Patients with radial ray deficiency often function surprisingly well, despite the radial deviation at the wrist and hand anomalies.³⁶ However, the extent of hand involvement has an impact on patients' functional ability.³⁸ These patients use an altered pattern of prehension that corresponds to their anatomic findings. Because structure and function of the radial digits are generally poorer than those of the ulnar digits, the radial deviation has some advantage. In this position, the ulnar border of the hand is optimally positioned for manipulation of objects.³³ Careful consideration of a child's pattern of prehension, specifically observing the preferred digits for function, is essential before making surgical recommendations. Centralization of the wrist of a child who prefers to use the ulnar digits may place the more functional digits at a less advantageous position for prehension.

Surgeons should recognize the difference between patients with thumb and hand anomalies associated with radial ray deficiency and those with isolated thumb hypoplasia. Manske et al⁴⁰ studied 23 patients who had pollicization surgery and compared usage of the hand between patients with isolated thumb hypoplasia and those with associated conditions. Eighteen patients had associated conditions, including radial ray deficiency, mirror hand, and five-fingered hand. The five patients with isolated thumb hypoplasia had an average of 95% usage of their hand with little difference between handling large and small objects. Patients with associated conditions used their hands an average of 79% of the time. These patients did not use their hand with the pollicized digit for a third of activities with small objects. Whether patients with associated anomalies will achieve an optimal functional outcome after reconstructive procedures needs to be considered.

Changes in the arterial system are also present. The brachial artery is usually normal but is sometimes divided into two branches high in the arm or not divided at all at the elbow.^{28a,36} The ulnar artery is usually normal in the forearm, but the radial artery is often small or absent.^{9,28a,36} The interosseous arteries are usually well developed and may accompany the median nerve through the forearm and supply the radial distribution in the forearm.^{9,28a,36}

Although rare, changes in the nerve supply may affect the limb as proximal as the root of the brachial plexus.³⁶ More frequently, the myocutaneous nerve is absent or abnormal and joined with or replaced by the median nerve.³⁶ The radial nerve terminates at the level of the lateral epicondyle.^{9,28a,36} If this occurs, the sensory distribution of the radial side of the hand is supplied by a larger superficial dorsal branch of the median nerve that emerges high in the forearm.^{9,28a} This nerve also communicates with the sensory branch of the ulnar nerve on the dorsal aspect of the hand.^{28a} Riordan^{28a} noted that the median nerve was the most superficial structure on the radial aspect on the forearm. The ulnar nerve was usually normal.^{9,28a,36}

Evaluation

Patients with radial ray deficiency should be evaluated shortly after birth. Documentation of associated conditions is important, especially in patients with thrombocytopenia-absent radius syndrome and those with cardiac anomalies.

The presence or absence of active elbow flexion in infants with Bayne and Klug type III or IV radial ray anomalies should be appreciated (see Table 42-1). This may be difficult to discern because of a child's developmental status and should be evaluated again at the child's next appointment. A musculoskeletal examination of the child's volar creases and flexion and extension digital creases provides information about the integrity of the joints. If creases are present, then the joint has been in use. Patients with Bayne and Klug type III or IV radial ray deficiency should be referred immediately to physical or occupational therapy for passive stretches, which are more easily accomplished if begun at an early age.

Functional evaluation of the upper limb can begin when a child is 4 to 6 months of age. As mentioned previously, the radial deviation of the wrist may place the more functional ulnar digits in a position for function. Centralization of such a hand may inhibit a child's function. When evaluating fine motor function, surgeons should consider the following four factors:

1. The presence of active elbow flexion. Absent or minimal active elbow flexion is a contraindication for centralization surgery.^{29,33,41,42}
2. A radial or an ulnar approach to an object during reach. If a child uses the ulnar side of the hand to manipulate objects, then centralization may not be of benefit. If the radial side of the hand is used, then the surgeon should determine whether the patient has an efficient gross and fine grasp. If a patient uses a pattern of opposition with the thumb and digits for fine and gross grasp, then centralization is recommended. If a patient uses an interdigital grasp between the index and long fingers, centralization followed by pollicization is a possibility. However, this recommendation is contraindicated for those with significant associated anomalies in the hand.^{29,41,42}
3. The child's development. If a child has a unilateral impairment, fine and gross grasp in the unaffected hand should be evaluated. Does the patient consistently demonstrate a superior pincer grasp with the unaffected hand? This information helps to determine whether a child's fine grasp has reached appropriate maturation. A key observation period for prehension is 9 to 12 months of age, when the infant develops a pincer grasp.⁴³
4. The child's suitability for postoperative rehabilitation. Will the child comply with the splinting and therapy regime? For an older child, has he or she adapted sufficiently to be independent in daily skills. Children older than 12 years of age have usually learned to adapt well, and surgery is rarely indicated.^{11,41}

Surgical correction often presents the conflict between the wish for cosmetic acceptance versus the risk of functional loss.¹¹ Several possible outcomes of centralization surgery may negatively influence function.³³ These include shortening the forearm from surgical trauma to the distal ulnar epiphyseal plate.^{33,39} After centralization of the wrist, function can be compromised from the inevitably associated stiffness.³³ Frankel et al⁴² did not recommend centralization for most patients with bilateral radial ray deficiency because of the possibility of functional loss. Although gains in function and appearance are reported in several studies, the authors indicated the difficulty in objectively measuring their outcomes.^{33,39,44} Comparing treated and untreated patients with radial ray deficiency is challenging.^{8,38} No standardized method exists for evaluating preoperative and postoperative function to validate surgical interventions in these patients.³⁸ Difficulties in measuring outcomes include rarity and heterogeneity of the condition, the presence of confounding associated anomalies, challenges in obtaining preoperative data because of the young age at which patients undergo surgical intervention, and finding untreated patients as controls.^{38,45} Furthermore, those who are untreated use adaptive techniques and aids (such as Velcro on clothing) and function well in their daily living skills.⁸ Most studies use additional components, such as ROM, sensation, strength, and prehension, as a means of measuring function, but only a few measure performance in daily activities.⁴⁶ Further research is needed to validate surgical outcomes in patients with radial ray deficiency.

Nonoperative Management

Splinting, serial casting, and passive stretches of the upper limb begin immediately after birth to maximize ROM.^{11,33} Thermoplastic static progressive splints are effective in providing a prolonged stretch to the hand and wrist on the forearm. If serial casts are used, they must extend above the elbow.¹¹ Splinting and casting should always be secondary to routine passive stretching exercise.¹¹ Passive stretching of the hand out of the flexed, pronated position with gentle axial traction and ulnar deviation is the priority.¹¹ Initially, parents may have difficulty applying traction to the limb,

but its importance should be emphasized, because it is a critical component of the stretch that positions the carpus over the head of the ulna. Stretches should be conducted daily at every diaper change (approximately eight times a day). Finger contractures and limitations in elbow flexion also require passive ROM stretches to optimize ROM.³³ We prefer to provide intensive passive stretching from birth. Splinting at night is introduced when children are approximately 3 months of age. At this age, the limb size facilitates a more optimal and effective stretch in a splint. Splints are not worn during the day to allow fine motor and sensory development. Effective splinting and stretching of the limb can prevent the need for soft tissue release in patients with Bayne and Klug type III and IV radial dysplasia before centralization or radialization.¹¹

Operative Management

Indications for surgical management vary by type of radial ray deficiency. For mild type I cases, operative treatment is rarely required. In some type I patients and mild type II patients, Ilizarov distraction has been demonstrated to successfully lengthen the radius. In severe type II, III, and IV patients, centralization is often indicated provided that active elbow flexion is present. Centralization has numerous contraindications. It should not be offered to adults who have adjusted to the deformity, because it may worsen function. Deficient or absent active elbow flexion is usually considered a contraindication to centralization. Tethering of neurovascular structures (for example, a radially displaced median nerve) may preclude centralization. Associated systemic disorders limiting lifespan are considered contraindications.

In appropriately selected patients, centralization produces a more normal-appearing, better-functioning limb. The components of centralization include a wide release of radially based contracted soft tissues and excision of any remaining fibrous anlage, transposition and temporary pin fixation of the hand onto the ulna, possible transfer of the extensor carpi radialis and flexor carpi radialis muscles to the ulnar side of the wrist, and local fasciocutaneous flap closure of the resulting radial-sided skin deficit (Fig. 42-9).

Preoperative management seeks to improve outcomes. Stretching, splinting, and serial casting reduce the likelihood that tight soft tissues will produce relapse postoperatively. In addition, if the soft tissues are adequately compliant, less bone needs to be resected from the carpus to accommodate the distal ulna, and in some cases, no bone needs to be resected from the carpus.

Some authors advocate using an Ilizarov distractor for soft tissue distraction before centralization. In principle, this can be considered a more invasive form of aggressive precentralization splinting. Nanchahal and Tonkin⁴⁷ recommended using distraction to straighten the ulna and improve the position of the hand before centralization. They thought that the procedure preserved carpal bones that would otherwise need to be resected in more difficult centralizations. They examined the child at 3 months of age, and if the hand could not be reduced onto the distal ulna, then they recommended soft tissue distraction. They combined an open soft tissue release with distractor application, if needed. After a latency period of 1 week, they distracted at 1 mm per day, usually for 3 to 4 weeks. Recently, however, Manske et al⁴⁸ compared the long-term outcomes of patients treated with centralization alone versus external distraction followed by centralization. They found that soft tissue distraction using an external fixator did not prevent recurrence of the deformity and was associated with worse radial deviation and volar subluxation of the wrist on radiographs.⁴⁸

Several different incisions and approaches have been reported for centralization and its variations. Evans et al⁴⁹ recommended using a dorsoulnar-based bilobed flap with a transverse radial incision. Manske et al⁵⁰ described a transverse ulnar approach. Buck-Gramcko⁵¹ reported an S-shaped incision from the dorsum of the hand to the proximal third of the forearm. A lazy-S incision was also used by Nanchahal and Tonkin⁴⁷, who excised excess skin and subcutaneous tissue

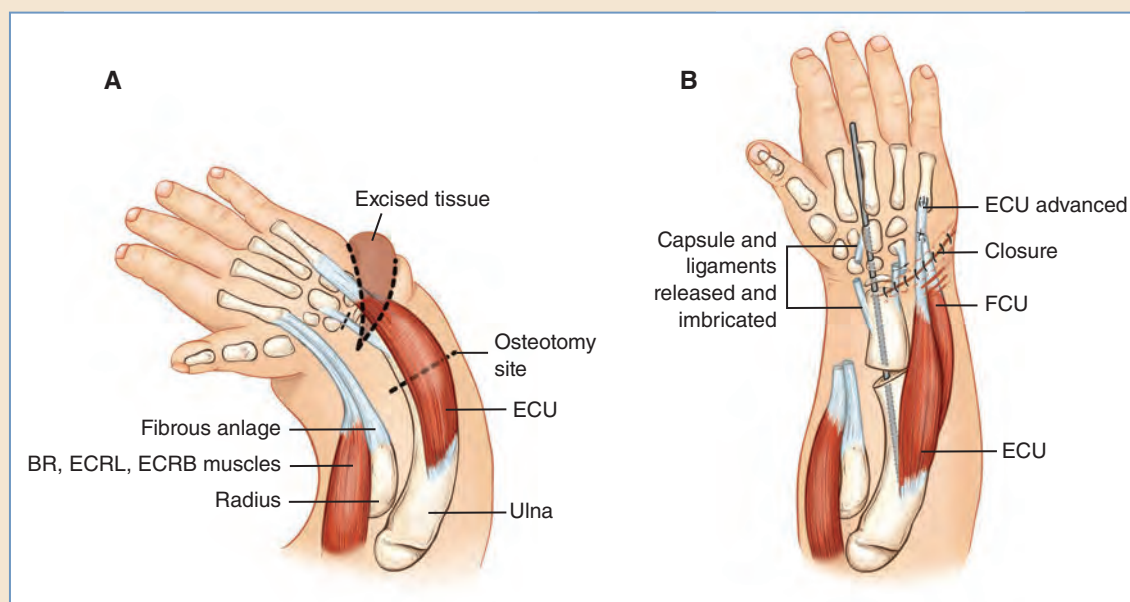


Fig. 42-9 In carefully selected patients, centralization can produce a more normal-appearing hand with improved function. **A**, Partial absence of the radius with a bowed ulna (Bayne and Klug type III radial ray deficiency). The radial-sided myotendinous units, including the brachioradialis, the extensor carpi radialis longus, and the extensor carpi radialis brevis, are hypoplastic. A tethering radial fibrous anlage is present. The radiocarpal joint capsule is tight. The median nerve is displaced radially and is located in an abnormally superficial plane. A radial-sided skin deficit and an ulnar skin excess may be present. **B**, The new anatomy after centralization. Exposure is carried out through an incision overlying the wrist that can be lengthened with a Z-plasty if needed. Alternatively, a bilobed flap can be raised for exposure and subsequent radial-sided skin coverage. The median nerve is dissected and preserved. The radiocarpal joint capsule is released. When possible, carpal bones are preserved. Sometimes it is necessary to remove a slot of bone and cartilage from the carpus to accommodate the distal ulna. The extensor carpi ulnaris is shortened and sutured to itself to generate a counterbalancing ulnar force across the wrist. An osteotomy of the radius is stabilized with a pin emerging through the dorsal wrist. Excess skin may need to be excised from the ulnar side of the wrist after centralization. (BR, Brachioradialis; ECRB, extensor carpi radialis brevis; ECRL, extensor carpi radialis longus; ECU, extensor carpi ulnaris; FCU, flexor carpi ulnaris.)

from the ulnar side of the wrist as necessary. Regardless of the approach used, full access to the ulna and the radial extensor and flexor tendons is needed. The decision to use a local flap, such as a bilobed flap, is based on whether a skin deficit exists when the hand is repositioned. If soft tissue distraction has been used, then a dorsally based flap may not be necessary, and a Z-plasty or lazy-S incision may be adequate.

After an incision is made, soft tissue is released. The median nerve (replacing the radial nerve) lies very superficial and radial to its normal location and can be easily damaged. The median artery may accompany it. It is usually not necessary to identify the ulnar artery, but it may be the sole arterial supply to hand. The extensor retinaculum is divided (except on the ulnar side), and the radial-sided wrist extensors and flexors are divided. In type III patients, the fibrous anlage should be excised. The ulnocarpal joint capsule is raised as a distally based flap, and the ulnar head is freed, with an effort to preserve the epiphyseal vascularity. The carpus is then placed onto the ulna. Lamb³³ and Manske et al⁵⁰ favored resection of the central carpal bones to accommodate the ulna, whereas Bayne and Klug²⁹ and Buck-Gramcko⁵¹ retained the carpal bones to maximize length. Buck-Gramcko⁵¹ placed the ulna in a more radial position (radialization) to shorten the level arm available to the radial-sided deforming forces that may produce relapse. He also trans-

ferred the extensor carpi radialis and the flexor carpi radialis to the ulnar side of the wrist for the same reason.⁵¹ Most authors shave a portion of the distal ulna to facilitate placement into the carpus, while preventing damage to the growth plate. An ulnar osteotomy is performed if indicated (for bowing or to decrease length to accommodate retaining carpal bones). A Steinmann pin or K-wire is placed from the index metacarpal into the ulna.

When possible, the radial wrist extensor and flexor mass is transposed to the ulnar wrist. This may not be possible if the myotendinous units are very short, fibrotic, or not present. Bayne and Klug²⁹ recommended transfer of the extensor carpi ulnaris and flexor carpi ulnaris distally to the base of the small finger metacarpal to act as a dynamic sling in an effort to minimize relapse. After the skin is closed, a plaster long-arm cast is applied. The pin is removed usually 6 weeks postoperatively or later. Pollicization is considered 6 months later.

Damore et al⁴⁵ presented five main surgical and nonsurgical factors for recurrence of deformity after centralization surgery:

1. The inability to obtain complete correction at surgery
2. Inadequate radial soft tissue release
3. Premature fixation removal
4. Poor splint use
5. The natural tendency for there to be radial deviation for functional use of the ulnar border of the hand

Postoperative splinting is essential for the success of centralization and radialization procedures.^{11,29,33,45} A thermoplastic, forearm-based wrist splint with the fingers and thumb free is fabricated after the fixation is removed. Damore et al⁴⁵ recommended gradual weaning of the splint to night use only and continued use until skeletal maturity. Bayne and Klug²⁹ recommended a more restrictive regime whereby the child is splinted during the day until 6 years of age and then weaned to night splinting. Lamb³³ recommended postoperative splinting for at least 6 months. We prefer a splinting protocol of a minimum of 6 months postoperatively and then night splinting until skeletal maturity (Fig. 42-10). When patients are transitioning to use a night splint, they should be gradually introduced to activities with greater resistance and force. Ongoing active rehabilitation is not required after this acute phase of recovery. However, therapists should continue to provide ongoing functional evaluations to optimize daily functioning.

Recurrent deformity is often reported after centralization procedures for radial longitudinal deficiency.^{48,52} Ulnocarpal arthrodesis has been proposed as a successful salvage procedure in children who developed recurrent radial deviation after centralization.⁵³ Other experienced surgeons have moved away from centralization procedures because of the associated loss of wrist motion,



Fig. 42-10 This 5-year-old child had a type III radial longitudinal deficiency and a Blauth type IIIb hypoplastic thumb on the left side and is shown after centralization and pollicization for thumb reconstruction (see Chapter 43).

the negative impact on ulnar growth, and the high recurrence rate of radial deviation.⁵⁴ These authors favored soft tissue-only procedures (for example, tendon transfers, release of tight structures, and local skin flaps) combined with long-term splinting of the wrist at night. Alternative proposed solutions to reduce recurrence of the radial deformity after centralization in patients with types III or IV radial longitudinal deficiency include vascularized second metatarsophalangeal joint transfer to the distal ulna.⁵⁵ Although this procedure is performed to improve carpal stability and the potential for physal growth, the technique is technically demanding and was associated with a high rate of complications.

In addition to centralization procedures, some authors have attempted to lengthen the ulna in patients with radial longitudinal deficiency to increase forearm length. Farr et al⁵⁶ reported an average increase in ulnar length of 75% in six patients after distraction lengthening, but corrections in ulnar bowing and radial deviation were not maintained at a mean follow-up of 4 years. Other authors are less enthusiastic about ulnar lengthening in radial longitudinal deficiency.^{57,58}

Central Deficiency: Cleft Hand and Symbrachydactyly

Barsky⁵⁹ subclassified central anomalies as typical cleft hand and atypical cleft hand. To clarify the classification of hand anomalies, the decision to discard the term *atypical cleft hand* was made at the Fifth Congress of the International Federation of Societies of Surgery of the Hand in Paris in 1992.⁶⁰ In the following discussion, the current terminology will be used: cleft hand (to include the former typical cleft hand) and symbrachydactyly (to include the former atypical cleft hand).

CLEFT HAND

In patients with a cleft hand, the cleft is usually along the middle finger ray, but clefting between the ring and little fingers with hypoplasia of the little finger may be seen.⁶¹ In a review of 11 patients with clefts between the ring and little fingers, the cleft reached only into the distal palm in most hands, but in two hands, the cleft extended to the proximal palm.⁶² In all patients the little finger was hypoplastic, and interphalangeal joint motion was limited. In severe cases of cleft hand, the suppression of digits is greatest on the radial side of the hand, whereas in symbrachydactyly, the ulnar side of the hand is most affected. According to Lister,⁶⁰ a hand with no wrist deformity and only a thumb shows symbrachydactyly, and a hand with only a little finger shows cleft hand.

Confusion in classification can easily arise, because clefting has been classified as a failure of formation, whereas the associated polydactylies are defined as a duplication of parts. This ought to make them independent entities. Manske⁶³ reported on identical twins with elements of both clefting and central polysyndactyly in the same hand or in different hands of the same individual. He suggested a possible continuum from central polysyndactyly with osseous fusion through the cleft hand with hypertrophic bony structures to cleft hand with an absent middle finger ray and no other bony changes.⁶³ Ogino⁶⁴ thought that cleft hand was pathogenetically related to syndactyly and central polydactyly and represented failure of the induction of finger rays. All of the variations seen will never be reconciled into a single category. We think that classifying cleft hand under failure of formation (longitudinal, central) is a simple, though neither elegant nor rigorous, solution.

Several classification systems have been developed to reconcile the wide variation seen in typical cleft hand patients.⁶⁴⁻⁶⁶ We prefer Blauth and Falliner's classification,⁶⁶ because it is easy to remember and suggests the degree of complexity that may be required in the reconstruction.

Table 42-3 Blauth and Falliner's Classification of Cleft Hand

Type 1	Cleft hand with osseous aplasia
Type 2	Cleft hand with synostosis
Type 3	Cleft hand with aplasia and synostosis

They analyzed both the literature and their own 35 patients. They emphasized the value of radiographs in assessing morphology and stated that, in 40% of their cases, they thought the cleft was caused by synostosis. Their classification system is presented in Table 42-3.

The right side is more commonly involved than the left, and most patients are male.^{5,66} Cleft hand may be familial¹⁰ (Fig. 42-11). The mode of hereditary transmission varies widely, but overall, the risk of inheritance of cleft hand was about 20% according to Birch-Jensen.¹⁰ Although most cases of cleft hand are sporadic, when hereditary, it is often dominant.¹⁰

Epidemiology

Birch-Jensen¹⁰ studied the incidence of cleft hand in the general population. In his report, the term *split hand* corresponded to typical cleft hand, and *symbrachydactyly* carried its current meaning. The cases of atypical split hand appear to belong to both current classes and cannot be reassigned retrospectively. For this reason, the birth data presented slightly underestimated the true incidence of these conditions. The stated frequency at birth for cleft hand was 1 in 90,000, and the prevalence in the population was 1 in 112,000. McGuirk et al¹⁴ reported the incidence of longitudinal arrest, central, as 0.06 in 1000 of all congenital upper limb malformations. More recently Ekblom et al⁵ reported that central longitudinal deficiencies occurred with a frequency of 0.9 per 10,000 live births. Cleft hand and symbrachydactyly accounted for 1.4% and 2.6%, respectively, of all congenital upper limb anomalies.⁵ Ogino et al³ reported that cleft hand was seen in 2.6% of children with hand anomalies and symbrachydactyly in 5.1% of cases if atypical cleft hand and brachysyndactyly were interpreted as symbrachydactyly.

The deformities can be broadly classified as longitudinal failures of formation of the central ray of the hand.¹ These lesions may result from defects in the undifferentiated mesenchyme occurring early in life in embryos at stages XX to XXII, as suggested by Senrui et al.⁶⁷ This is supported by the finding that the vascular anomalies found in congenital hand lesions either by dissection⁶⁸ or at angiography⁶¹ correspond to an arrest in the development of the vascular tree at an appropriate early stage.⁶⁸ Surface markings can also suggest possible mechanisms involved in these deficiencies. For example, whorls are not normally seen in the interdigital areas of the distal palm in normal hands. Whorls are seen in cleft hands on the ulnar side of the cleft over the top of the rudimentary middle ray,⁶⁹ suggesting an association with, and confirming the suppression of, the central ray.

When other associated anomalies are present, inheritance patterns are usually autosomal dominant, as seen in cleft hand with major defects of the ulna, radius, clavicle, acetabulum, femur, tibia, or fibula; absent permanent teeth; nasolacrimal duct obstruction; eye defects; and onychia.⁷⁰ The ectrodactyly (congenital partial or complete absence of one or more digits), ectodermal dysplasia, and cleft lip and palate (EEC) syndrome is inherited in an autosomal dominant fashion, but about half of the cases are sporadic,⁷⁰ and great variability of expression is seen.⁷¹

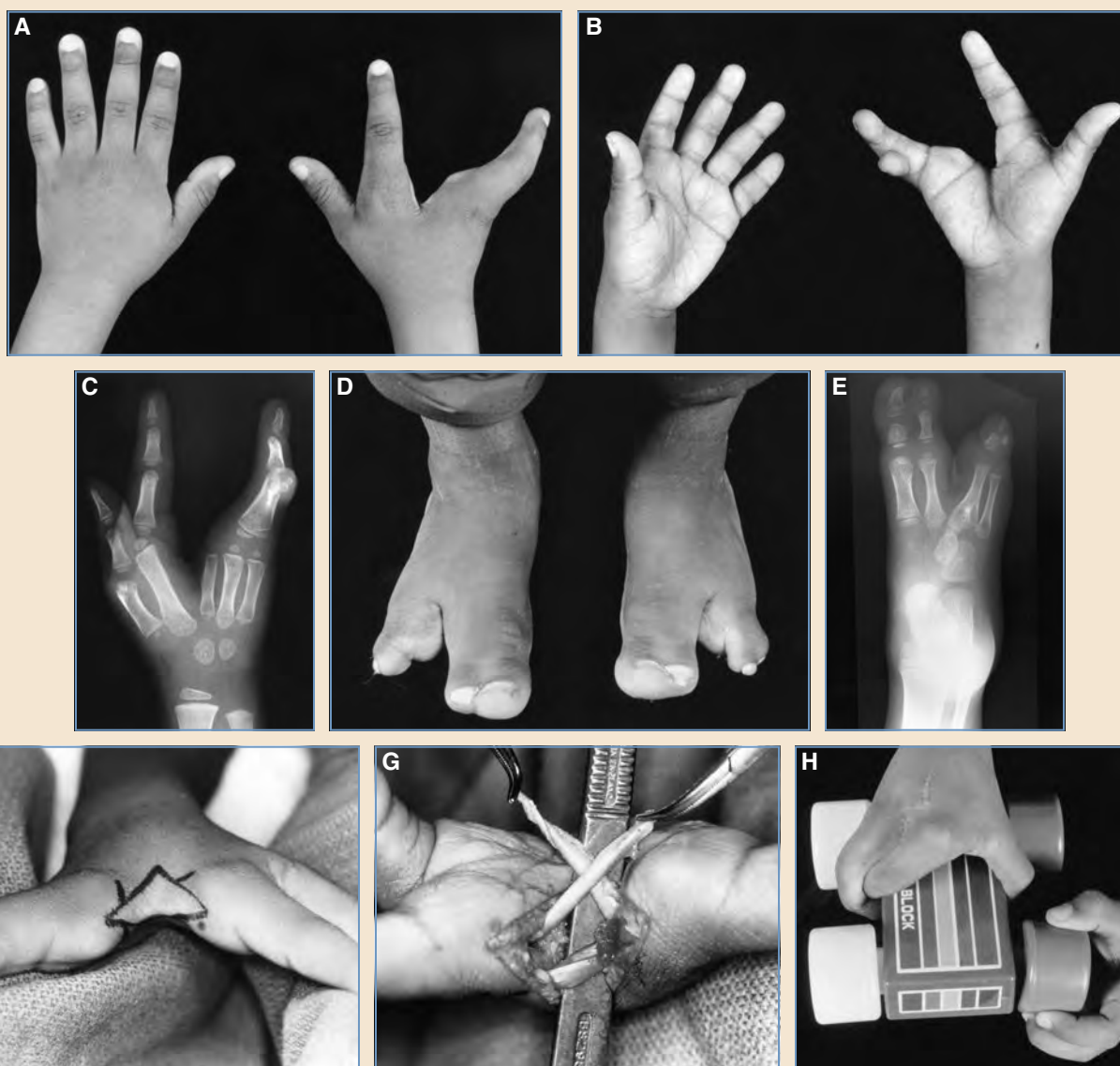


Fig. 42-11 **A**, Although only one hand is involved, this patient has many features of typical cleft hand. A deep cleft is present in the center of the hand, and adjacent digits are relatively preserved. **B**, The volar view shows that the central cleft is actually wider than the first web space. The ring and little fingers are fused. **C**, A radiograph of the hand shows an unsatisfied third metacarpal, fused proximal phalanges of the fourth and fifth digits, a small distal nubbin for the little finger, and a tight first web space. **D**, Bilateral foot anomalies are seen with suppression of the central ray, soft tissue clefting, and well-proportioned but syndactylyzed border digits. **E**, Radiography of the right foot shows fusion of the third and fourth metacarpals, absence of the third toe, and a soft tissue cleft. The first surgical step in hand reconstruction in this patient was to ensure that grasp between the thumb and index was sufficient. When the patient was 5 years of age, a four-flap Z-plasty of the first web space was undertaken, along with reconstruction of the ulnar collateral ligament of the metacarpophalangeal joint of the thumb with fascia lata. **F**, After ensuring in therapy that the patient could transfer gross grasp to the reconstructed first web space, the redundant tissue of the cleft was excised, Z-plasties were performed along the incision lines, and the central cleft was closed in a simple fashion when the patient was 5½ years of age. **G**, A long segment of fascia lata was harvested and used to reconstruct the deep transverse metacarpal ligament. The fascia was passed in a figure-of-eight fashion around the necks of the metacarpals of the index and ring fingers and secured with a nonabsorbable suture. **H**, The patient has good hand function after cleft closure, because the function of the first web space was assessed, reconstructed, and rehabilitated before cleft closure was undertaken.

Lower extremity deformities (see Fig. 42-11), such as clefting, syndactyly, polydactyly, and tibial defects, occur with cleft hand.^{64,66} Patients with foot anomalies may have a deep, V-shaped cleft of the central hand, often with syndactyly of the remaining components, and do not have intercalary defects.⁵⁹ Syndromic associations in cleft hand are not common but must be considered because of their implications for overall treatment. Rüdiger et al⁷² first described the association of ectrodactyly, ectodermal dysplasia, and cleft lip and palate. Patients with the EEC syndrome can have significant problems during or after anesthesia, including difficulties with malnutrition, difficulty controlling body temperature (related to hypohidrosis), and persistent urinary tract infections.⁷³ Even more rare is the association of ectodermal dysplasia, ectrodactyly, and macular dystrophy (EEM syndrome) reported in an isolated community in Japan.⁷⁴ Cleft hand has occurred in patients with Silver-Russell syndrome, a form of dwarfism of prenatal onset.⁷⁵

Anatomy

Carpal anomalies are not seen in patients with a distal or partial cleft of the metacarpals.⁷⁶ Patients with complete defects of the metacarpals have defects confined to the distal carpal row. These include hypoplasia of the trapezoid, enlargement of the hamate, and splitting of the capitate. Fusion of the second metacarpal to the trapezoid, fusion of the capitate to the hamate, and fusion of the first metacarpal to the trapezium occurs.⁷⁶

Cleft hand may be seen on one side, with syndactyly or central polydactyly on the opposite side.⁶⁴ Associated anomalies usually include syndactyly of the thumb and index finger (Fig. 42-12) and syndactyly of the ring and little fingers. Cleft hand can be pathogenetically related to syndactyly and central polydactyly with failure of induction of finger rays, and triphalangeal thumb, shortened middle phalanx of the little finger, transverse phalanges, double metacarpals supporting a single digit (see Fig. 42-11), and combined metacarpals have been reported.⁶⁴

Nonoperative Management

Treatment for cleft hand varies widely depending on the presentation. Many patients present with little or no functional impairment, and operative treatment may be indicated only for cosmesis rather than functional gain.^{59,77} The inability to improve function in some cases was recognized even in the nineteenth century.⁷⁸ Evaluating function in patients with cleft hands before surgical treatment is critical, because prehension may be good and cosmesis the only issue (Fig. 42-13). Gross grasp within the cleft may be much more efficient than grasp between the thumb and index finger, and function may be compromised if the cleft is closed. Therefore a thorough evaluation of prehension and bimanual function is required before reconstruction is considered. For example, Ogino⁶⁴ recommended ablation of the interdigital cleft in patients with no missing finger or one missing finger. He did not recommend cleft closure in patients missing the index finger or the index and middle fingers and in patients missing three digits, because the ability to grasp large objects will be diminished.

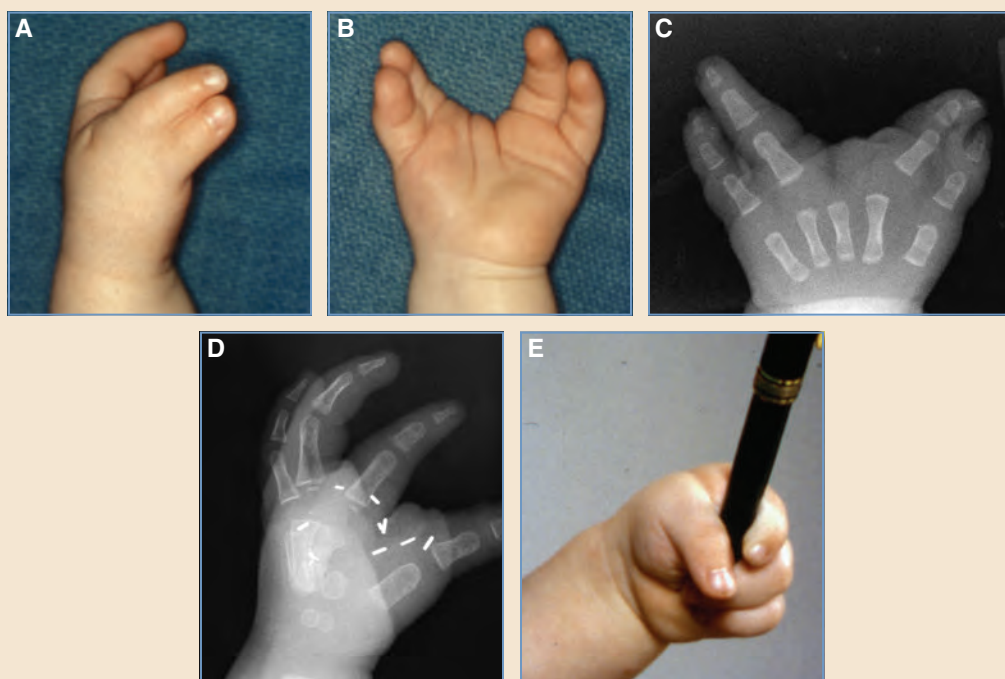


Fig. 42-12 **A**, This 1-year-old patient had a cleft hand with absence of the middle finger and thumb-index syndactyly. **B**, A gross grasp was present in the central cleft. The tight, incomplete simple syndactyly between the thumb and index finger produced significant radial clinodactyly of the index finger. **C**, A preoperative radiograph showed very tight skeletal alignment of the first and second rays. The middle finger metacarpal was unsatisfied, and a wide, central soft tissue cleft was present. **D**, This oblique postoperative radiograph shows that the newly created first web space is wide, and that the second metacarpal is now unsatisfied, because the index finger was moved onto the stump of the middle metacarpal. **E**, The patient is able to grasp small and larger objects. Further correction of the radial clinodactyly of the index finger may be required when the patient is older.

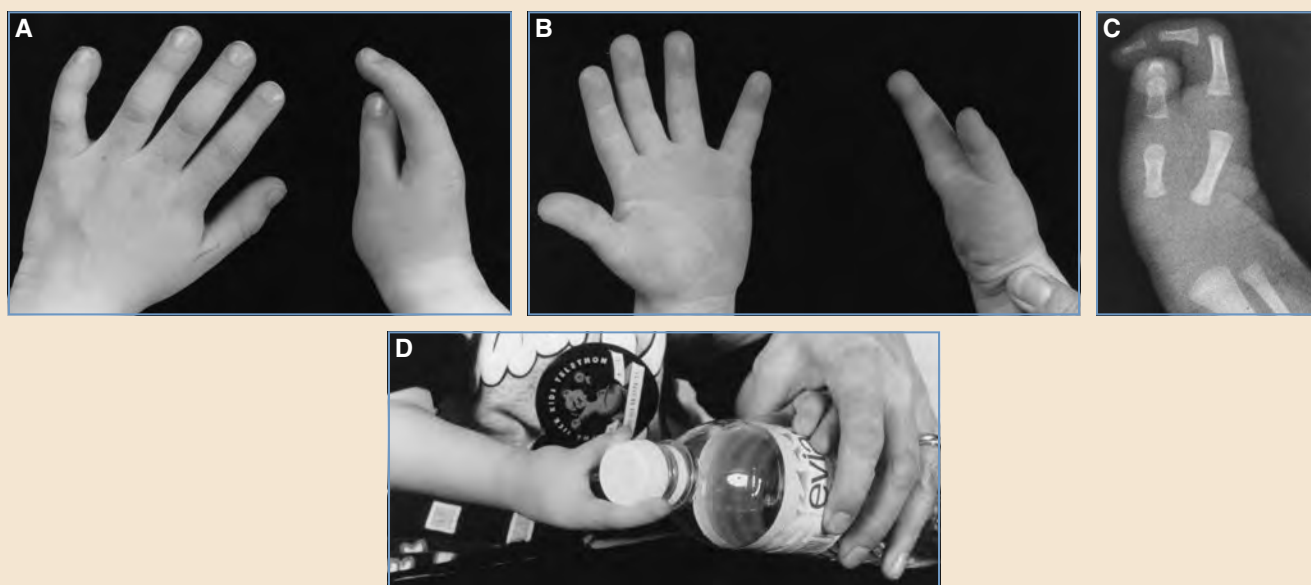


Fig. 42-13 **A** and **B**, A two-fingered cleft hand with a sufficient web between the digits may prove very functional and not warrant reconstruction. **C**, A radiograph shows the two digits positioned in an excellent orientation for grasp. **D**, Hand function could not be improved with surgery.

Operative Management

Upton and Taghnia⁷⁷ summarized the key indications for operative intervention for cleft hand. These included severe flexion contracture of one or more digits, a malpositioned index ray that interferes with function, a tight syndactyly involving the thumb, and major functional limitations of the hand. Furthermore, they described the following reconstructive goals:

1. Preservation or reconstruction of a mobile, unscarred, and stable thumb
2. Transposition of the index ray to the ulnar side of the cleft
3. Creation of a wide, well-contoured first web space
4. Correction of index malrotation or deviation
5. Preservation of an intact (if present) adductor pollicis muscle
6. Creation of a satisfactory appearance

Timing of Operative Management

A tight thumb to index syndactyly should be released as soon as possible.⁶⁴ However, the child must be old enough to demonstrate the functional patterns that will be used in prehension. Generally, the following procedures involving flap transposition are best performed when patients are about 2 years of age, when the hand is larger than in early infancy, and the child has sufficiently sophisticated function to make use of the reconstruction. Ligamentous reconstruction is easier at this age. Rotation osteotomies can be readily performed in patients 5 to 6 years of age, when the skeleton is more mature.

Surgical Technique

Closure of the soft tissue cleft should achieve a smooth surface on the palm and dorsum of the hand and should produce a new central web that is natural in contour and correctly positioned. Barsky⁵⁹ described a hexagonal flap, based distally on one side of the cleft, to reconstruct the interdigital web (Fig. 42-14). He closed the cleft proximally with interdigitating, zigzag incisions. Kelikian and Doumanian⁷⁹ described treating cleft hand patients with a rectangular flap with its pedicle based at a level of the normal commissure, if one exists on the opposite hand. Ogino⁶⁴ preferred a small triangular flap, usually based on the ulnar side of the cleft, which could be used to break up the scar along the commissure. Finally, in patients with a generous amount of avail-

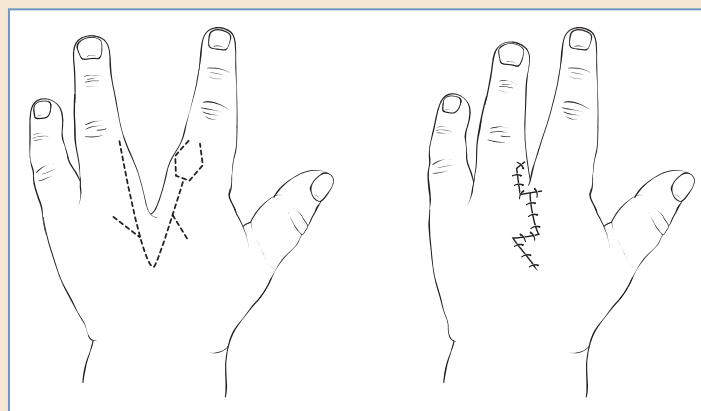


Fig. 42-14 Barsky⁵⁹ introduced a distally based hexagonal flap for reconstruction of the commissure in cleft hands. Proximal zigzag incisions are used in the closure.

able soft tissue, direct closure of the web may be possible using Z-plasties along the length of the volar and dorsal incisions to reduce the risk of scar contracture (see Fig. 42-11).

Various techniques have been used to reconstruct the deep transverse metacarpal ligament, one of the key steps to successful cleft hand reconstruction. Barsky⁵⁹ used drill holes and heavy gut sutures to oppose the metacarpal heads and removed unsatisfied metacarpals in the cleft. Ogino⁶⁴ and Tsuge and Watari⁸⁰ used ligamentous flaps designed on the adjacent sides of the flexor sheaths. Usually the A1 pulley is divided longitudinally at its attachment opposite the cleft and transposed across the cleft to be sutured to a corresponding flap from the other side (Fig. 42-15). This allows a reconstruction at the appropriate anatomic site, without excessive tension. Fascia lata is available in most patients and provides a ready source for ligament reconstruction (see Fig. 42-11). It is wound around the necks of the metacarpals in a figure-of-eight fashion to provide stable fixation of the cleft. Finally, an infrequently available but efficient option for donor material for reconstruction of the deep transverse metacarpal ligament is the use of ligaments or tendons from a part that is being discarded, such as the Achilles tendon from an amputated hypoplastic leg (Fig. 42-16). An intertendinous connection can be made between the extensor tendons of the digits on either side of the cleft to prevent future separation.⁸⁰

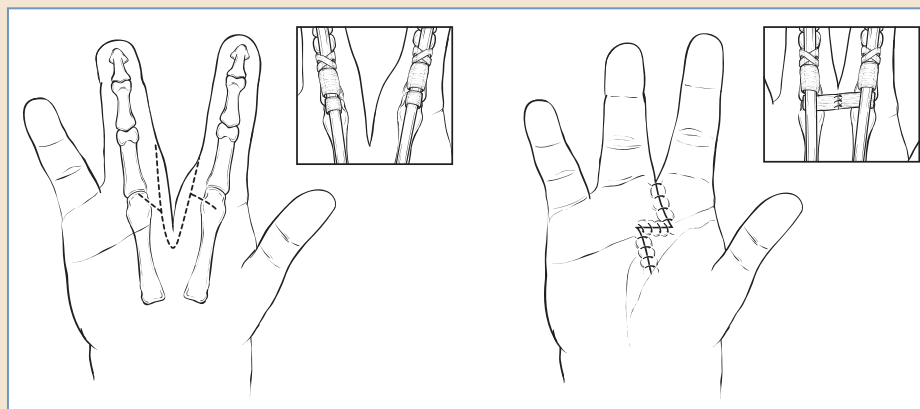


Fig. 42-15 Flexor tendon sheath, usually the A1 pulley, can be used to reconstruct the deep transverse metacarpal ligament. The sheaths are incised on the side opposite the cleft and then are transposed and sutured to each other.



Fig. 42-16 This hypoplastic leg (failure of formation of the tibia) had been amputated through the knee to allow fitting of a prosthesis. The Achilles tendon (held above the forceps) was dissected from the specimen and used for reconstruction of the deep transverse metacarpal ligament in a cleft hand. The extreme hypoplasia of the tendon suited the size of the desired reconstruction.

Ray fusion or osseous syndactyly is frequently seen in patients with cleft hand. Tsuge and Watari⁸⁰ attempted to separate these if the shape of the phalanges could be preserved. Angiography helped to evaluate for a common arterial supply, but the primary difficulty was the shared tendons and joint capsule. An interesting finding from their work was that the extensor tendons tend to fuse more proximally than the flexors. The gains from these extensive procedures may, however, be modest.

Syndactyly of the thumb and index finger is often seen (see Fig. 42-12), and the treatment for the central cleft must be planned with this in mind. One of the most satisfying reconstructions for patients with a large central cleft and complete syndactyly between the thumb and index finger is the Snow-Littler procedure.⁸¹ The digital portion of the syndactyly between the thumb and index finger is divided as for a typical syndactyly, with zigzag incisions and full-thickness skin grafts. A flap based on the palm is taken from the central cleft space on a neurovascular bundle and is used to resurface the newly created first web space (Fig. 42-17). Lateral pressure is placed to oppose the rays adjacent to the cleft to evaluate the amount of skin that can be resected in the area of the cleft.⁶⁴ The dorsal veins are preserved with the flap. All fibrous bands are released between the thumb and index finger. The venous drainage of the index finger is preserved by a dorsal skin bridge. The index finger is transposed on its neurovascular bundles⁸² at the level of the metacarpal base to close the cleft. The base of the middle metacarpal can be used for fusion to the base of the index (see Fig. 42-12), sometimes in the form of a bony peg. If the middle metacarpal is

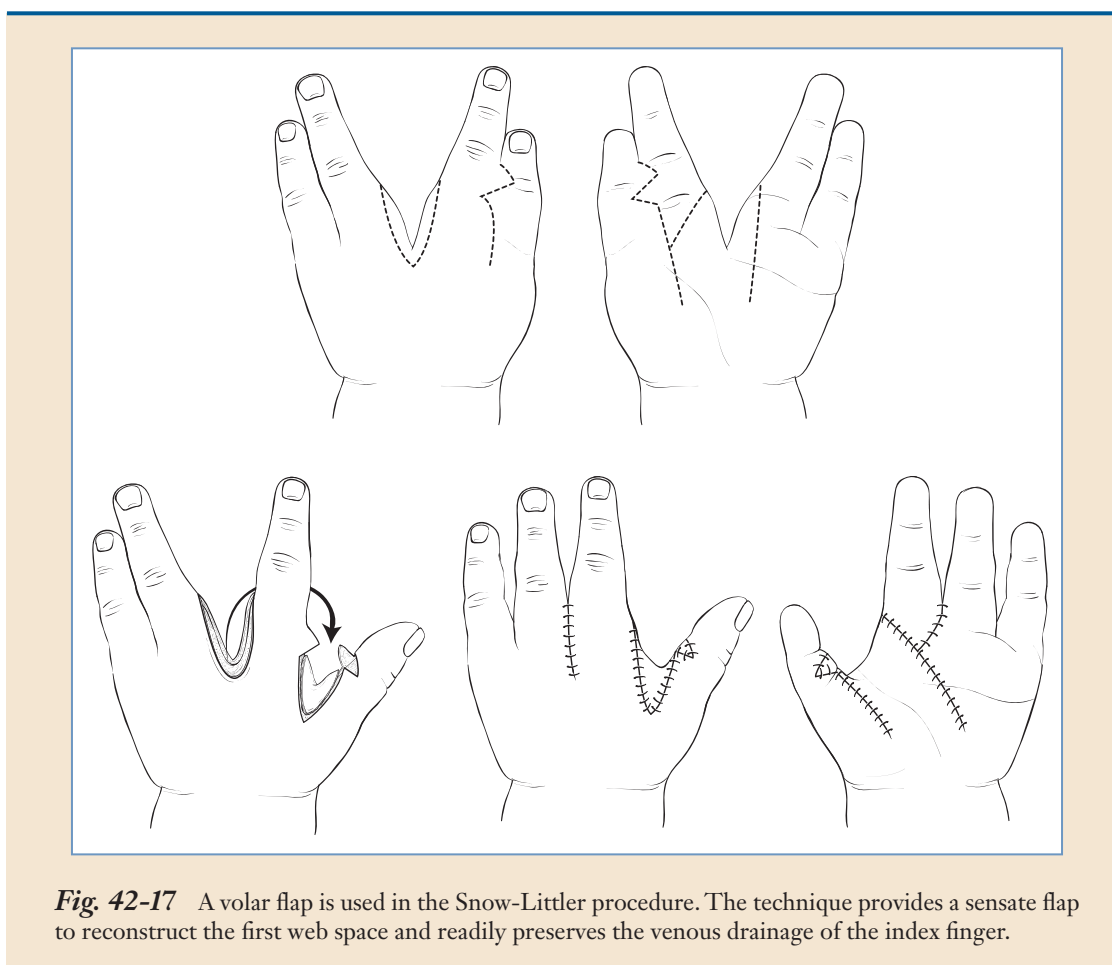


Fig. 42-17 A volar flap is used in the Snow-Littler procedure. The technique provides a sensate flap to reconstruct the first web space and readily preserves the venous drainage of the index finger.

tiny or absent, transposition may not be possible. In such a case, it may be necessary to undertake a wedge osteotomy of the index finger to bring it into alignment without transposition. Proper alignment of the metacarpals is critical.⁸¹ Transverse metacarpals or phalanges that impede closure of the second and fourth metacarpals should be resected and the extensor hood reconstructed as necessary.⁶⁴ When transverse metacarpals are significant contributors to a joint, a portion of the bone may need to be preserved to retain stability of the joint.⁸⁰ Patients can have very good function after this procedure, with good grasp of small and larger objects (see Fig. 42-12).

Miura and Komada⁸³ designed an operation with simpler incisions than those used in the Snow-Littler operation for reconstruction of the first web space in cleft hand patients (Fig. 42-18). Incisions are made along the edge of the cleft and around the dorsal base of the index finger. The skin originally proximal to the incision on the index is used to close the new first web space. Although this is less demanding than the Snow-Littler operation,⁸¹ it probably will not provide sufficient tissue for release of a very tight thumb.

Another option for treating clefts with partial syndactyly of the thumb and index finger is a dorsally based flap, as described by Ogino⁶⁴ (Fig. 42-19). The excess volar skin is outlined in a zigzag fashion, an ulnar triangular flap is used to reconstruct the central web, and the incision is carried dorsally between the fingers adjacent to the cleft. A volar incision to open the first web

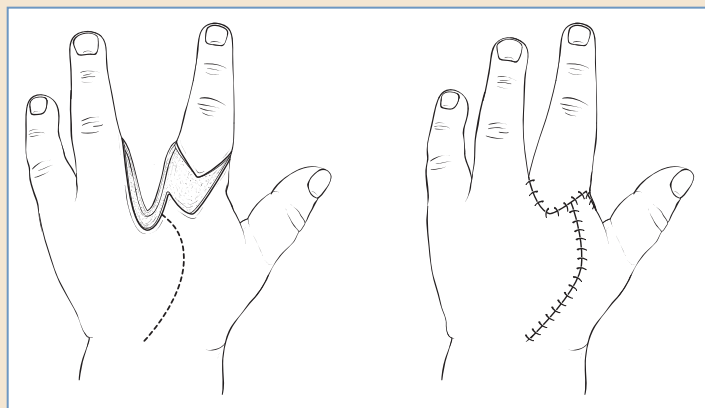


Fig. 42-18 Miura's procedure provides a simple reconstruction in a cleft hand patient with an adducted thumb but provides only a limited amount of tissue for web reconstruction. It is not useful in patients with syndactyly of the first web space.

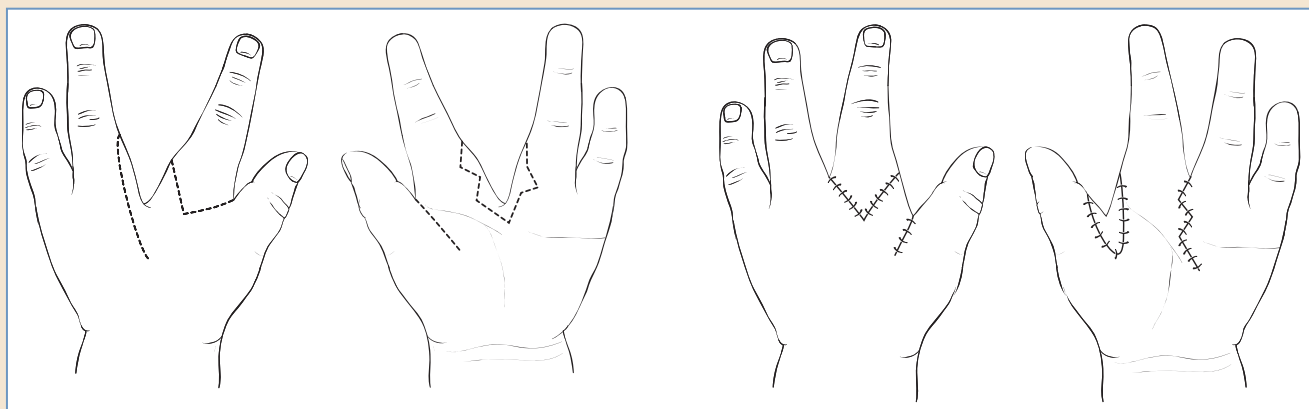


Fig. 42-19 Ogino used a dorsally based flap to reconstruct the tight first web space in cleft hands with partial syndactyly of the thumb and index finger. The venous drainage of the index finger must be carefully preserved in this operation.

space is extended dorsally to a V-shaped apex over the dorsum of the index finger and is carried distally along the ulnar border of the index finger to meet the volar excision in the cleft. The dorsal flap is transposed to the dorsal aspect of the first web space after all the appropriate structures are divided. Additional full-thickness skin grafts may be required.

In some patients with syndactyly between the ring and little fingers, this may be released at the same time as the cleft closure.⁶⁴ Any flexion deformity of the proximal interphalangeal joint of the ring finger may not need to be corrected if the cleft surgery is performed before the patient is 1 year of age. If correction of the flexion deformity is necessary, the superficialis tendon is released from its insertion and secured to the base of the proximal phalanx volar to the reconstructed transverse metacarpal ligament. If the fingers adjacent to the cleft are divergent, the two slips of the superficialis tendon can be split and directed to the bases of the proximal phalanges of the fingers on either side of the cleft to help actively reduce the deviation.⁶⁴

A rotational osteotomy of the border digits can be performed, often at a later stage, to bring them into better alignment for grasp. They should be aligned so that the tips of the digits pass each other in flexion to allow a stronger power grasp.⁶⁰

Patients with clefting between the ring and little fingers may require surgery for cosmesis but not for function.⁶² Flaps, as described by Barsky,⁵⁹ have been used to line the web. Generally, the little finger does not tend to drift away, and little is required to maintain the metacarpal alignment.⁶²

Few studies document the outcomes of operative intervention for patients with cleft hand. In 1984 Tada⁸⁴ published his results of surgical treatment of 31 hands with central ray deficiency and reported satisfactory function and aesthetic outcomes. In 2008 Goldfarb et al⁸⁵ evaluated outcomes of bony and/or soft tissue closure of clefts in 12 patients (16 hands) using a subjective appearance-related visual analog scale completed by the parent and surgeon, ROM measurements, and radiographic divergence angles of the index and ring fingers. The authors noted improvement in all assessed outcomes at a minimum 5-year follow-up.⁸⁵ In a later study by Aleem et al,⁸⁶ narrowing of the first web space and index finger metacarpophalangeal joint abnormalities were associated with worse functional outcomes after surgical reconstruction of the cleft hand, whereas the presence of a transverse-lying metacarpal within the cleft was not predictive of outcome.⁸⁶ Further studies using well-defined and validated outcome measures are needed to better define outcomes of treatment for cleft hand, to refine operative indications, and to identify areas for improvement in perioperative management.

Authors' Preferences

Careful planning is the key factor for a successful procedure, and a preoperative evaluation of the existing prehension patterns cannot be emphasized too strongly (see Fig. 42-11). The greatest pitfall in reconstructing cleft hands is to close the cleft when function cannot be transferred to the first web, thereby losing gross grasp.

We prefer the Snow-Littler procedure⁸¹ in clefts with thumb-index syndactyly, because the volarly based flap is extremely robust and remains sensate. Any of the flaps described previously are satisfactory for closure of the web itself; however, the reconstruction of the deep transverse metacarpal ligament must be tight and the skin closure loose. The simplest option for reconstruction of the deep transverse metacarpal ligament is a transposed A1 pulley.

SYMBRACHYDACTYLY

Symbrachydactyly (Fig. 42-20) shows more severe suppression of hand formation, with attenuation of the marginal rays and central fingers sometimes represented by nubbins (Fig. 42-21).

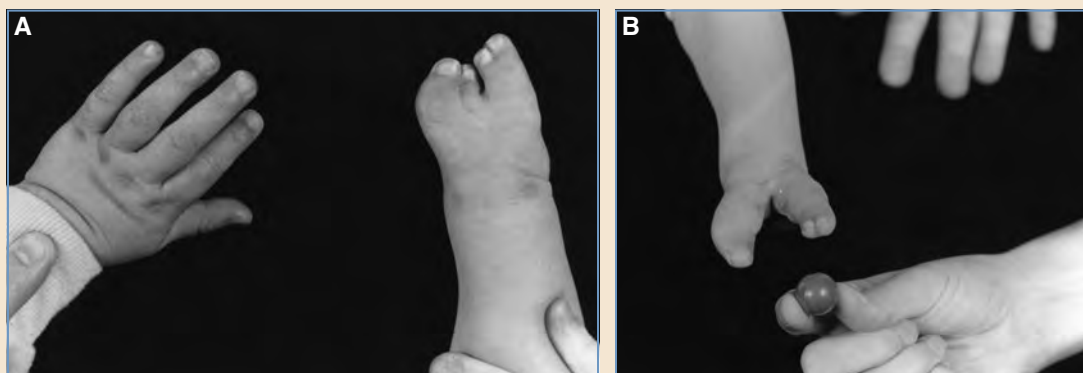


Fig. 42-20 **A**, This 18-month-old child had symbrachydactyly of the right hand. No digital motion was observed, but some independent activity of the border metacarpals was noted. No grasp was present. The surgical plan was to discard the third ray, which was extremely rudimentary, and open a cleft to the level of the proximal metacarpals. Zigzag incisions were employed both dorsally and volarly to reduce the risk of contracture along the cleft (see Fig. 42-23). The unsatisfied raw areas were covered with full-thickness skin grafts. **B**, Postoperatively, the child uses the hand for simple grasp and should be able to use it to assist the master hand in light tasks.

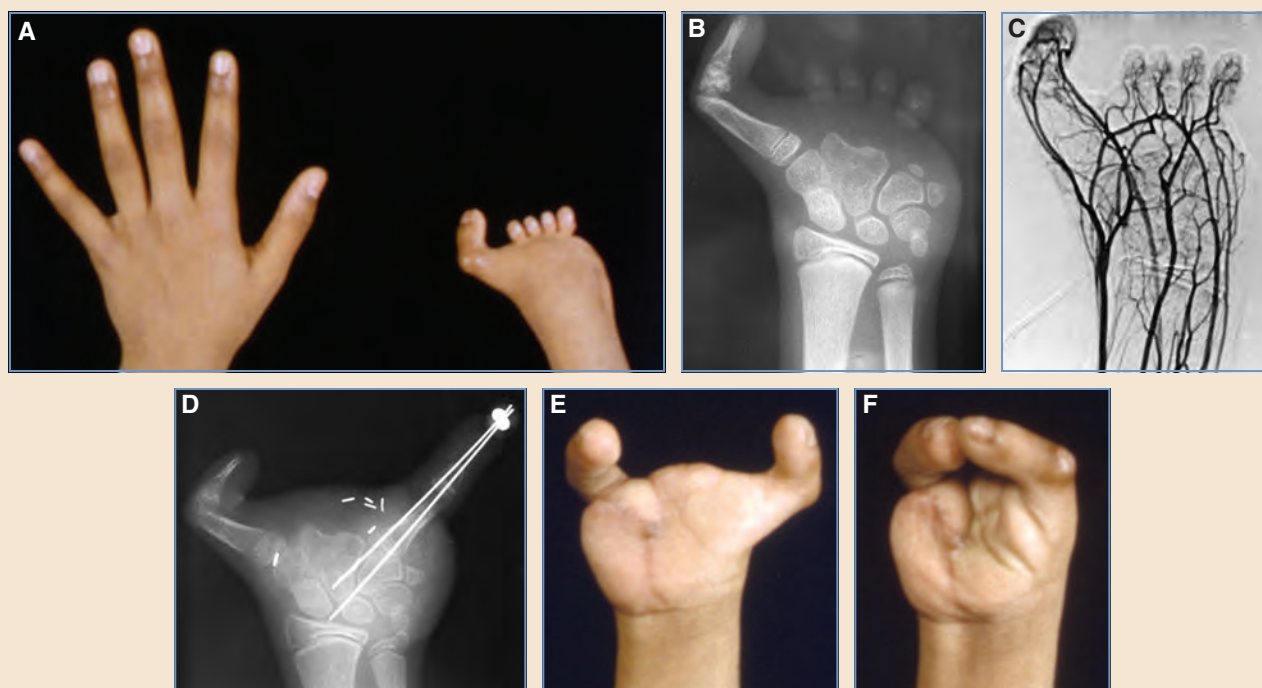


Fig. 42-21 **A**, This 12-year-old's right hand had symbrachydactyly with a poor thumb and only nubbins representing the rest of the fingers. The patient was only able to hold very small objects in this hand. A two-stage reconstruction was performed. The first stage involved the thumb, and the second involved the ulnar digit. **B**, The lax thumb was reconstructed using an intercalary bone graft of fibular hemicortex, shown in a radiograph, to stabilize the thumb. A passive prosthesis was fitted on the ulnar side of the hand with a methylmethacrylate model of the proposed toe transfer. This prosthesis provided an opportunity to test various potential positions for the new digit. **C**, An AP arteriogram of the hand revealed an intact deep volar arch and a persistent median artery. A large ulnar artery was seen and was the planned vessel for arterial reconstruction. **D**, The second toe was transferred to the ulnar hand and held in place with two K-wires. The cartilage was shaved from the opposed surfaces at the osteosynthesis site. **E**, A postoperative volar view shows a second toe-to-ulnar hand transfer in place. The maximum extension is shown. It allows gross grasp sufficient for moderately sized objects. **F**, Pulp-to-pulp pinch is achieved mostly with thumb and metacarpal movement, with little movement in the transferred toe.

Table 42-4 Blauth and Gekeler's Symbrachydactyly Classification

Type	Characteristics
Type 1	Short finger type with short or absent middle phalanges and incomplete syndactyly
Type 2	Cleft hand with absence of one or more central fingers
Type 3	Monodactyly with a hypoplastic thumb and missing the remaining digits except for nail-bearing remnants
Type 4	Peromelia with absence of all digits except for small humps bearing nail fragments

Characteristically, symbrachydactyly includes rudimentary nails with very hypoplastic digits, whereas cleft hands have no rudimentary digits.⁶⁰

Blauth and Gekeler⁸⁷ attempted to classify symbrachydactyly (Table 42-4). Their paper was based on 19 of their own patients and 179 from the literature.

Symbrachydactyly is usually unilateral¹⁰ and without foot involvement or family history.⁵⁹

Epidemiology

In Birch-Jensen's study¹⁰ the incidence at birth was 1 in 40,000, and the prevalence in the population was 1 in 47,000. These patients may have deformities of the lower extremities, particularly clubfoot. Most cases of symbrachydactyly were sporadic; in the few hereditary cases, it appeared to be recessive in nature.

Anatomy

Most patients with symbrachydactyly have generalized hypoplasia of the carpal bones.⁷⁶ In some patients it is associated with pectoral hypoplasia and is a common feature in Poland syndrome.⁶⁴

Associated Conditions: Poland Syndrome

The original description of Poland syndrome was that of George Elt, a 27-year-old convict whose cadaver was dissected by Alfred Poland⁸⁸ in 1840 at Guy's Hospital in London. In 1921 Pol⁸⁹ published the first extensive description of a series of 20 cases involving pectoral muscle defects and hand deformities. He also noted 13 cases with isolated symbrachydactyly. The typical hand deformity seen in Poland syndrome is symbrachydactyly with absent or short middle phalanges,⁹⁰ syndactyly, and a normal or small thumb (Fig. 42-22). This corresponded well with Poland's original description. Surgical findings in the hand may include a common trunk forming the superficial or deep flexors and adhesion of the flexors to the surrounding tissues.⁶⁷

Many additional findings have been associated with Poland syndrome, including hypoplasia of the breast and nipple-areola complex,^{91,92} rib involvement,^{93,94} Pierre Robin sequence,⁹⁵ Moebius syndrome,^{93,96} dextrocardia,^{94,96} and morning glory syndrome, which features an unusual appearance of the optic disc, with a coloboma, and usually blindness thought to result from disturbances in closure of the optic disc.⁹⁷ Other associations include undescended testis, clubfoot, and cleft palate.⁹³

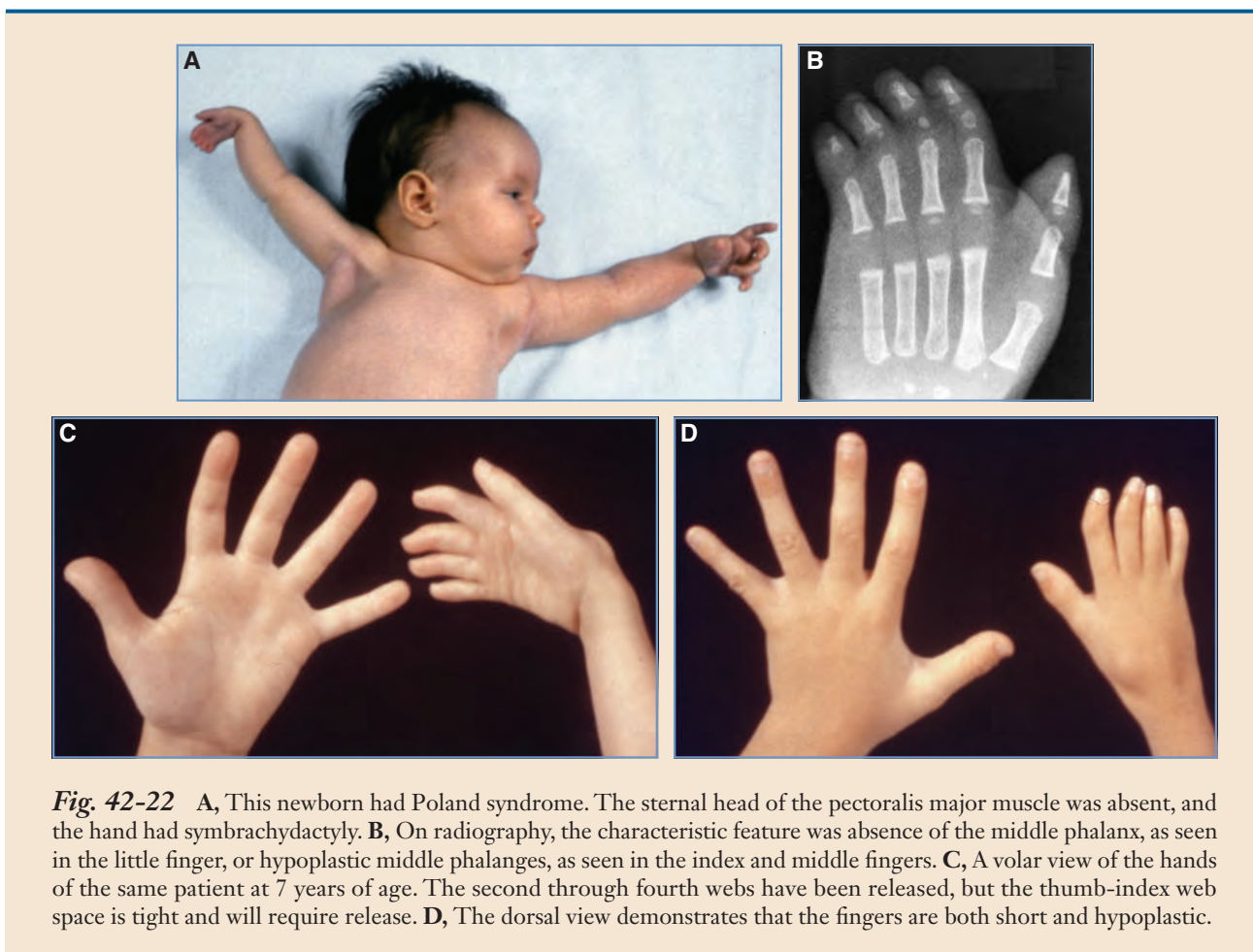


Fig. 42-22 A, This newborn had Poland syndrome. The sternal head of the pectoralis major muscle was absent, and the hand had symbrachydactyly. B, On radiography, the characteristic feature was absence of the middle phalanx, as seen in the little finger, or hypoplastic middle phalanges, as seen in the index and middle fingers. C, A volar view of the hands of the same patient at 7 years of age. The second through fourth webs have been released, but the thumb-index web space is tight and will require release. D, The dorsal view demonstrates that the fingers are both short and hypoplastic.

Epidemiology of Poland Syndrome

McGillivray and Lowry⁹³ examined 44 patients. A family history was obtained in 28 of them. The sternal head of the pectoralis major was absent in all patients, and symbrachydactyly was present in most. All family histories were negative. The authors found that the overall incidence was 1 in 32,000 live births in British Columbia, Canada. The prevalence in the population in 1975 was 1 in 57,000. Castilla et al⁹⁸ reviewed 599,109 live births in South America from 1967 to 1977. The overall incidence of complete Poland anomaly in South America was 1 in 50,000. They agreed with the Canadian study in showing a higher incidence in males, consistent unilaterality, preferential right-sidedness, no familial cases, and a history of prenatal drug exposure in a third of patients studied.

An autosomal dominant form of inheritance has been suggested, but with few positive family histories, the data are inconclusive for a familial cause for Poland syndrome.^{70,91,92} Engber⁹⁹ thought that cleft hand (as opposed to symbrachydactyly) in association with pectoral aplasia was an entity separate from Poland syndrome. It was a sporadic anomaly and families can be counseled accordingly.

Evaluation

Examining the function of a patient's hand deficiency is essential. In particular, the patterns of manipulation of small and large objects and of bimanual hand function will indicate the patient's preferred method of grasp. It is critical to determine whether grasp in a central cleft is the most efficient available. This clinical information can be supplemented by a radiologic assessment of the part with attention paid to the presence of hidden polydactylies and skeletal impediments to useful function.

Nonoperative Management

In some patients, surgery cannot produce functional or aesthetic improvement. Children should be followed every other year. A watchful follow-up helps to ensure the child and family that a team is in place, has an interest in their condition, and will keep them informed of advances as new concepts arise.

Operative Management

A preoperative evaluation of symbrachydactyly patients is critical to successful surgical management. Surgeons should determine whether the existing digits on the hand are functional, particularly if adduction of the border metacarpals is strong and purposeful. Are the border digits useful as they are, or can they be rendered sufficiently strong with a bony or ligamentous reconstruction or even with a tendon transfer? All patients with symbrachydactyly should initially be considered as candidates for toe-to-hand transfer if only to eliminate the possibility. Some of the procedures described in the following sections use toe parts as donor tissue. These must be undertaken with caution to preserve the option of whole toe transfer in appropriate patients.

Timing of Operative Management

Simple soft tissue procedures such as opening a cleft to enhance grasp can be performed as soon as a child demonstrates purposeful grasp with the opposite hand, at about 6 months of age. Non-vascularized toe phalanx transfers should be carried out before patients are 15 months of age. Free vascularized toes can be transferred to hypoplastic hands in patients as young as 1 year of age in some centers. We think this is best performed when patients are about 4 years of age, when the vascular structures are more robust and children may be better able to cooperate in therapy.

Surgical Technique

Patients with symbrachydactyly, no functioning digits, and significant hypoplasia may benefit from the creation of a cleft in the hand (see Fig. 42-20). A central ray or rays that are extremely hypoplastic can be discarded and a cleft opened to allow lateral pinch between the remaining radial and ulnar rays. Zigzag incisions, as used in syndactyly releases, and full-thickness skin grafts are required (Fig. 42-23). This may provide a simple helper hand when adequate proximal tissues for the addition of one or more toe transfers will probably not be available.

Patients with active digits that are too short for good function may benefit from nonvascularized toe proximal phalanx transfer, first described for this purpose by Carroll and Green.¹⁰⁰ These patients attempt to use the available digits for grasp, but the digits are of inadequate length to be useful. Preoperative strength and ROM in the digits to be reconstructed must be good. Among the large series describing this procedure, Buck-Gramcko and Pereira¹⁰¹ provided a most detailed

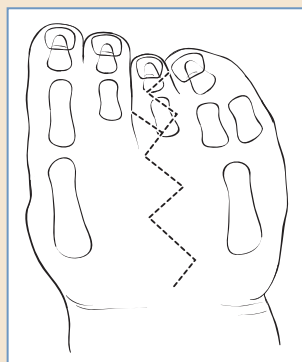


Fig. 42-23 An approach analogous to that used in syndactyly surgery is used in opening a cleft in symbrachydactyly. Zigzag incisions are planned on the volar and dorsal surfaces and interdigitate after release of the cleft. Hypoplastic central digits may need to be sacrificed in opening up the cleft. Raw surfaces are covered with full-thickness skin grafts (see Fig. 42-20 for clinical results).

description of the technique and a history of the procedure. It can be used to lengthen partially aplastic digits containing no proximal phalanx and to provide a hemiarthroplasty by retaining the plantar plate and collateral ligaments on the transplanted toe phalanx. The dissection of the toe proximal phalanx is started distally by opening the proximal interphalangeal joint. Proximally, the plantar plate of the metatarsophalangeal joint is divided at its proximal border if it is to be used for joint reconstruction. The periosteum of the phalanx must not be damaged; it is removed intact with the toe phalanx and improves the probability of successful revascularization. After the toe is removed, the extensor and flexor tendons are sewn together with nonabsorbable sutures to reduce the final shortening of the toe and are sewn proximally to the collateral ligament remnants to reduce lateral drift. If the toe phalanx is secured with an arthrodesis, the articular surface of the base of the proximal phalanx of the toe is shaved off, and the epiphysis is preserved. If a joint is reconstructed, the plantar plate is left free and not sutured.¹⁰¹

In preparing the hand, Goldberg and Watson¹⁰² used a zigzag incision over the dorsum of the finger. The skin incision did not violate the tip of the dissected pouch, reducing the risk of extrusion. The deeper soft tissue overlying the end of the stump was incised in a volar-dorsal direction, and the tissue was spread to produce attachments for the collateral ligaments. A longitudinal K-wire was placed for fixation.

Buck-Gramcko and Pereira¹⁰¹ reviewed 46 patients with 69 transferred phalanges and a mean follow-up of 40 months. The amount of postoperative growth was limited, but growth was better if the toe phalanx was transplanted in patients younger than 18 months of age. In the joint reconstruction patients, no factors correlated with the amount of motion achieved, and ROM was limited. They thought that hand function was improved, although this was not documented. Lister¹⁰³ thought that the insertion of nonvascularized toe phalanges might provide useful length in a much simpler operation than toe-to-hand transfer, if performed before patients are 15 months of age. Goldberg and Watson¹⁰² have shown that the percentage of open epiphyses at follow-up was greater in infants than in older children. In some cases, further distraction lengthening of the transplanted phalanx can be undertaken.¹⁰¹

Garagnani et al¹⁰⁴ recently reviewed long-term donor-site morbidity after nonvascularized toe phalangeal transfer in 40 children treated for digital hypoplasia. They concluded that donor-site complications were common, including donor toe instability, hypoplasia of adjacent phalanges, visible deformity that worsened with growth, difficulties with footwear, and emotional problems

related foot appearance.¹⁰⁴ These results highlighted that donor morbidity should not be overlooked in the preoperative discussion for nonvascularized toe phalanx transfers.

Another option is segmental transposition to augment a digit. Dobyns¹⁰⁵ defined this as the transfer in the same hand of a significant segment of a digit with one or more neurovascular pedicles to another digit to augment its size or length. He considered this particularly useful in small hands with a short thumb and index. Transposing the index remnant onto the thumb and discarding the proximal index ray provided a longer thumb and a deeper first web space.

Authors' Preferences

In deciding on the surgical treatment of symbrachydactyly, our first consideration is the preoperative function of the existing hand. For hands that are extremely rudimentary with poorly formed metacarpals and little intrinsic muscle function, the creation of a cleft in the hand may allow light grasp. Stronger hands, particularly those with a functional or reconstructible thumb, will often benefit from toe-to-hand transfer. Only hands with good individual finger and thumb function and a rudimentary grasp will benefit from a nonvascularized toe phalanx transfer.

We think that the gains from whole toe transfer in most cases of symbrachydactyly outweigh the gains from nonvascularized toe phalanx transfer. The key factor is to determine whether the hand is sufficiently developed to allow reconstruction of the existing digits or to support toe transfer, or, if the hand is extremely hypoplastic, whether to open a cleft in the center to facilitate simple side-to-side pinch. Ablation of the nubbins should generally be left to the discretion of the child. In all of these patients, the family should be prepared to expect a helper hand rather than a hand with subtle dexterity.

Future Considerations

Despite the ongoing research into the underlying causes and manifestations of these congenital differences of the hand, future management will probably change either as a result of a better understanding of the genetics involved or as a result of technical advances. Genetic developments may lead to early diagnosis and potential elimination of these defects.¹⁰ We may look forward to a time when rejection reactions are sufficiently controlled to allow elective hand transplantation without the need for long-term immunotherapy.

New techniques untried by most hand surgeons may lead to new avenues of therapy. For example, Ipsen and Barfred¹⁰⁶ described an unusual and complex reconstruction for cleft hand. They reported the case of a 12-year-old patient with two digits: a broad, large radial digit and an unstable, small ulnar digit. The ulnar digit did not have a carpometacarpal joint and was not animated. A new basal joint for the ulnar digit was created using a Swanson arthroplasty, and the flexor carpi ulnaris, extensor carpi ulnaris, and extensor digitorum communis tendons were transferred to the ulnar digit for animation. An additional tendon transfer to the radial digit was later required to provide good adduction. Key pinch was achieved. The use of the Swanson arthroplasty in young children to reconstruct a previously nonexistent joint is not yet a standard technique and must be considered with caution, because the durability of these prostheses over many decades of hard use has not been demonstrated. Long-term follow-up will be required to fully evaluate this procedure.

Other technical advances may facilitate our treatment of these unusual and challenging anomalies. We must remember, however, that function is crucial, and that no operation, however fascinating and elegant technically, is worthwhile if the child does not make use of the reconstruction.

ULNAR RAY DEFICIENCY: POSTAXIAL ANOMALY

Ulnar ray deficiency is the absence or hypoplasia of the ulna. It is generally characterized by a degree of limitation in the elbow joint, a shortened forearm, an ulnarly deviated wrist, and two or three radial digits in the hand. However, its clinical presentation is more variable than radial ray deficiency.^{11,34,107} The deformities in the elbow, wrist, and hand are highly variable.¹⁰⁸ The degree of carpal or ulnar deficiency has no apparent relationship to the position of the hand on the forearm.⁹ Hand anomalies can vary, with deficits in the radial, central, and ulnar digits with no specific pattern.⁹

Several classifications have been developed for ulnar ray deficiency.¹⁰⁸⁻¹¹² The classification from Dobyns et al⁹ (known as *Bayne's classification*) combines several other works and incorporates both elbow and ulna characteristics. It is considered to be the most clinically useful (Table 42-5).¹¹

Cole and Manske¹¹⁰ evaluated 45 limbs with ulnar ray deficiency and observed that 73% had abnormalities in the thumb and first web space. Twenty-eight of the 53 surgical procedures recommended were targeted at improving function in these two anatomic structures. They developed the following classification according to the thumb and first web that may assist with surgical recommendations (Table 42-6).

Table 42-5 Bayne's Classification of Ulnar Ray Deficiency

Type	Characteristics
I Hypoplasia of the ulna	Presence of distal and proximal ulnar epiphysis
II Partial aplasia of the ulna	Presence of the distal or middle third of the ulna
III Total aplasia of the ulna	Complete absence of the ulna
IV Radiohumeral synostosis	Fusion of the radius to the humerus

Table 42-6 Cole and Manske's Classification of Thumb and First Web Space in Ulnar Ray Deficiency

Type	Characteristics
Type A	The thumb and first web are normal.
Type B	The first web has mild deficiency, mild thumb hypoplasia, intact extrinsic tendon function, and opposition function.
Type C	The thumb has varying degrees of hypoplasia, and the first web has moderate to severe deficiency, including thumb-index syndactyly, often associated with malrotation of the thumb into the plane of the other digits, loss of opposition, and dysfunction of the extrinsic tendons.
Type D	The thumb is absent.

Epidemiology

Absence or hypoplasia of the ulna is one of the rarest congenital malformations of the upper extremity.^{107,113} In a recent study, Ekblom et al⁵ documented the incidence of ulnar dysplasia as 1 in 10,000 live births, accounting for 4.3 % of all congenital limb anomalies. McGuirk et al¹⁴ reported a lower incidence of 0.08 in 1000 among patients with congenital upper extremity malformations. Most cases are sporadic^{8,15,34,112} with no patterns of family inheritance.^{3,15,114} However, inheritance of three consecutive generations was reported.¹¹⁵ A simple pattern of inheritance is indicated in a small subgroup of these patients.¹⁵ The role of genetic and environmental factors in the cause of ulnar ray deficiency is not well understood.^{9,15}

Ulnar ray deficiency occurs bilaterally in approximately one of every three cases.^{110,112,116} The incidence is slightly increased in males with a ratio of 3:2^{107,112} or greater.^{5,110} Generally, there is no difference in right to left incidence.^{5,107,108,110,112}

The three most common syndromes related to ulnar ray deficiency are Cornelia de Lange syndrome, femur-fibula-ulna syndrome (or proximal focal femoral deficiency [PFFD]), and Schinzel syndrome.¹⁵ The last two syndromes are very rare. Swanson et al¹¹² found associated anomalies in the lower extremity in 41.5% of patients. Other associated conditions include the following:

- Contralateral hand defects¹¹⁶
- Clubfoot or associated anomalies of the lower extremity^{108,109,112,114}
- Scoliosis^{108,112,116}
- Spina bifida¹¹⁴
- Congenital dislocation of the hip^{107,109,114}
- Congenital coxa vara^{108,112}
- Oral facial malformations^{15,112,116}

Anatomy

Anatomic manifestations of ulnar ray deficiency are highly variable^{112,116} (Table 42-7). The previously mentioned classification systems were developed to provide a framework for understanding the anatomy of the forearm and hand. No correlation has been noted between the severity of ulnar deficiency, elbow deformity, and the degree of ulnar deviation at the radiocarpal joint.¹¹⁶ The proximal structures can also be involved, including hypoplasia of the shoulder structures, shortened humerus, and failure of differentiation of the elbow joint.⁹

Unlike radial ray deficiency, ulnar deficiencies most frequently present as partial absences.⁹ The articulation of the remaining portion of the ulna to the humerus is generally reasonably good,¹¹³ but radial head dislocation^{113,116} or radioulnar synostosis^{9,116} may occur. The ulna may be ankylosed to the humerus.^{8,9} Flexion deformities at the elbow often occur when the ulna is absent.⁹ In older children, restriction in elbow range is usually the result of radial head dislocation.⁹

Patients can compensate for restricted elbow and forearm movement by using shoulder and wrist movement.^{107,109} If functional deficits persist, surgical management of the humerus or forearm may optimize positioning of the upper extremity for function.^{107,109} Elbow flexion contractures may also pose difficulties with dressing activities. A surgical release may enhance function.¹⁰⁷

Ulnar bowing of the radius occurs in the forearm.^{8,116} The ulna is short, and the absent distal ulna may be replaced by a fibrocartilaginous anlage.¹¹³ This band of tissue restricts longitudinal growth,¹¹⁷ thereby restricting wrist motion and possibly contributing to the bowing of the radius.⁸

Table 42-7 Commonly Absent or Abnormal Anatomy in Ulnar Ray Deficiency

Structure	Commonly Absent	Abnormal
Bone	Ulnar carpal bones Ulnar metacarpals and digits	Shoulder girdle Humerus Ulna Radius Carpal bones Digits Thumb Triceps
Muscle	Triceps EDC of D5 EDM Extensor carpi ulnaris Palmaris longus FDP of D5 FCU	
Nerve	Superficial radial nerve at the level of the lateral epicondyle	

This table is by no means exhaustive, and many variations are known to occur.^{8,11,110,114}

D5, Fifth digit; EDC, extensor digitorum communis; EDM, extensor digiti minimi; FCU, flexor carpi ulnaris; FDP, flexor digitorum profundus.

The wrist is generally slender because of the absence of the ulnar carpal bones.⁹ The remaining carpal bones articulate with the distal radius.¹¹³ Broudy and Smith¹¹⁶ observed that the carpal bones were always affected with a common pattern of absence of the carpal bones proximal to any missing digital rays.¹¹⁶ Radial deviation at the wrist was decreased in all patients. They also observed a delay in appearance of carpal ossification centers in 18 of 26 limbs studied.

Less than a third of patients have fixed ulnar deviation at the wrist, and most are able to bring their wrist and hands into neutral position.⁸ Ulnar deviation at the wrist is generally nonprogressive and does not cause significant functional difficulties.¹¹⁶ Significant ulnar deviation can be prevented by early surgical management, and late or persistent deformity can be managed with an osteotomy if functional difficulties arise.

Ectrodactyly is the most common deformity in the hand.¹¹⁶ The remaining digits may show some degree of syndactyly.^{112,116} A correlation between the type or degree of ulnar deficiency and the location or number of absent digits has not been found.¹¹⁶ However, ulnar digits are absent more often than radial digits.¹¹⁶

Mild to moderate hypoplasia typically occurs in the thumb.¹¹⁶ Cole and Manske¹¹⁰ found thumb and first web space deficiencies in 73 % of the cases studied (see Table 42-6).

The radial digits are often preserved in patients with ulnar ray deficiency.¹¹⁶ Function is good in patients with opposition between the thumb and the radial digits. They are able to manage dressing, toileting, eating, and writing activities.¹¹⁸ Otherwise, reconstructive procedures for the thumb and web spaces may enhance opposition and prehension.¹¹⁶ Patients without opposition

will have impaired prehension. Adaptive prehension, such as an interdigital grasp and techniques or aids, may be used to facilitate independence. Despite altered prehension, most patients do not have difficulties in their activities of daily living.¹¹⁸

Evaluation

Children with ulnar ray deficiency should be initially evaluated shortly after birth. Evaluating wrist deviation and finger anomalies, such as camptodactyly and syndactyly, is important at the initial visit. Patients with contractures are referred for occupational and/or physical therapy. Early detection of contractures in the wrist and fingers can be managed through stretching or splinting to maximize ROM.

Nonoperative Management

The rehabilitation of infants with ulnar ray deficiency is focused on maximizing ROM and preventing contractures in the available joints. Passive ROM stretches of ulnar deviation contractures at the wrist and camptodactyly of the fingers are initiated as early as possible. Patients can wear a thermoplastic forearm-based wrist, hand, and finger splint at night to stretch the wrist into neutral alignment. This splint can be beneficial for children with camptodactyly to provide a prolonged stretch of the proximal interphalangeal joint into extension. Periods of stretching and splinting may be required throughout childhood to maintain optimal function. Caregivers should be advised on how to monitor their child's ROM.

Caregivers are instructed to promote bimanual activity to encourage their child to optimize the use of the affected limb. Therapists may suggest specific toys and activities to promote bimanual skills. For children beginning school, therapists assist with the acquisition of specific daily living skills such as learning to zip up a coat. As patients mature and develop specific hobbies and interests, therapists may assist in making these activities feasible, for example, by providing prosthetic adaptations for sporting equipment and musical instruments (see Fig. 42-4).

Operative Management

Ulnar ray deficiency can present quite variably. Therefore indications for operative management should be considered relative and not absolute; an individualized treatment plan should be stressed. Clinical outcome studies have been very limited because of the rarity of the condition. Elements of ulnar ray deficiency that may limit function and are often amenable to correction or improvement include syndactyly, first web space contracture, thumb duplication, thumb hypoplasia or malposition, restriction of growth with bowing of the radius from a tethering ulnar fibrous anlage, elbow stiffness, and elbow instability.

Syndactyly may be simple or complex. Standard release techniques work well for these deformities in many patients. However, for those with severe ectrodactyly, regional or distant flaps may need to be introduced to further separate the digits to allow a pincer grasp. First web space

contractures can be released with a Z-plasty or with other local flaps. The thumb ray may be present but poorly positioned, with the thumb pad in the same plane as the other digits. In such cases, the thumb may be rotated into a more advantageous position, allowing pulp-to-pulp pinch.

Carroll and Bowers,¹⁰⁷ Ogden et al,¹⁰⁸ and Broudy and Smith¹¹⁶ recommended excision of the fibrous ulnar anlage to prevent growth restriction and exacerbation of radial bowing. In patients with an unstable elbow and chronic dislocation of the radial head, excision of the proximal radius may be combined with excision of the distal ulna to form a one-bone forearm. Although the child can no longer pronate or supinate postoperatively, the elbow usually is stable.

Miller et al¹⁰⁹ recommended rotational humeral osteotomy in patients whose involved hand is positioned on the flank or buttock at rest and who cannot effectively place the hand in a functional position in front of the body. The affected forearm is also frequently malrotated; however, forearm osteotomies are rarely needed to improve function.⁵⁴

LONGITUDINAL INTERSEGMENTAL DEFICIENCY

From 1958 to 1962, the incidence of children with longitudinal intersegmental deficiency related to the ingestion of the sedative thalidomide by expectant mothers increased.^{119,120} These children typically presented with absence or deficiency in the bones between the hand and shoulder. Their hands or rudimentary fingers arose directly from the distal end of the remaining shoulder¹²⁰ (Fig. 42-24).

This condition was initially defined as intercalary deficiency, because the middle portion of the limb was absent or deficient, but the proximal and distal portions were present.¹²¹ The existence of true intercalary deficiencies was later questioned.¹ Although the condition affects mainly the long bones in the limb, the distal portion is never truly normal.^{1,122} Therefore these conditions are classified as a variation of longitudinal failure of formation of parts.¹



Fig. 42-24 A radiograph of a newborn with longitudinal intersegmental deficiency.

Table 42-8 Frantz and O’Rahilly’s Classification of Longitudinal Intersegmental Deficiency

Type	Characteristics
1: Complete	Absence of the proximal portion of the upper limb Hand attached to the trunk
2: Proximal	Absence of the humerus Forearm bone between the hand and trunk
3: Distal	Distal absence of the radius and ulna Hand attached to the humerus

Table 42-9 Tytherleigh-Strong and Hooper’s Classification of Longitudinal Intersegmental Deficiency

Type	Characteristics
A	An abnormal humerus with an abnormal single forearm bone
B	An abnormal humerus with an abnormal radius and ulna
C	An abnormal humerus fused to a forearm bone or bones

Attempts to further classify the conditions are reported. Frantz and O’Rahilly¹²¹ developed a classification system that consisted of three subtypes (Table 42-8). More recently, Tytherleigh-Strong and Hooper¹²² demonstrated that not all patients with longitudinal intersegmental deficiency could be classified under this system. Their classification system is shown in Table 42-9.

Epidemiology

Longitudinal intersegmental deficiencies account for 0.8% of congenital upper limb malformations.³² McGuirk et al¹⁴ reported an incidence of 0.06 in 1000. This condition is generally sporadic¹²¹ and can be induced by the use of thalidomide during pregnancy.¹¹⁹ A variety of cardiac malformations are associated with longitudinal intersegmental deficiencies.¹²⁰ Other associated conditions include Robert syndrome, cleft lip and/or palate, Holt-Oram syndrome, and adactyly-adontia syndrome.¹⁵

The pathogenesis of longitudinal intersegmental deficiencies is not completely known.¹²³ Thalidomide decreases growth in the limb-bud mesenchyme.¹²⁴ Tabin¹²⁵ proposed a theory using the progress-zone model to explain the pattern of perseveration of distal structures. According to the theory, thalidomide exposure causes the apical ectodermal ridge to produce fibroblast growth factor, which leads to respecification of cells in the limb bud. Others disagree that the apical ectodermal ridge plays a role because of the timing of limb development and thalidomide exposure.^{123,126} Last, evidence of involvement of the sensory nervous system in the pathogenesis of the limb deficiency has been shown.¹²³

Anatomy

Longitudinal intersegmental deficiencies are usually bilateral.³² Tytherleigh-Strong and Hooper¹²² studied 44 patients. Twenty patients were affected bilaterally, and just more than half had symmetrical deficiencies. Frantz and O'Rahilly,¹²¹ however, found that bilateral skeletal deficiencies are usually not symmetrical. In a cohort of patients affected by thalidomide in West Germany, half of the sample had only upper limb deficiencies. The other patients had a variation of upper and lower limb, ear, and other malformations.¹²⁰ Other studies of patients affected by thalidomide demonstrated that upper limb deficiencies were characteristically on the radial side.^{123,127} Variations of deficiency in the hand have a varying number of digits.¹²¹

Patients who are affected bilaterally without lower extremity involvement, often learn to become adept in using their feet for function and dexterity.²¹ Typically, functional problems are toileting and feeding activities. Therapy involving the use of adaptive devices and training is required to facilitate independence in daily living. Those who are affected unilaterally can use their affected hand with or without prostheses to assist their dominant unaffected hand to perform bilateral activities.

Evaluation

Children with longitudinal intersegmental deficiency should be initially seen within the first 3 months after birth. Those with joint contractures that require passive ROM exercises or splinting are referred for rehabilitation. Therapy is initiated at this time to work with the child and family to promote developmental milestones and to facilitate use of the lower extremities for children with bilateral involvement.

Nonoperative Management

Joint contractures such as camptodactyly are managed with passive stretches to maximize joint range. Thermoplastic splints may be required in severe cases.

Patients with longitudinal intersegmental deficiencies may be candidates for prosthetic management (see Fig. 42-4).²¹ Control of a terminal device can be designed using finger access, depending on the length of a patient's limb. The challenge in designing prostheses for these patients is to produce a simple and lightweight device; otherwise, compliance with the device will be low.²¹

Operative Management

There are few indications for operative management in patients with longitudinal intersegmental deficiency. Overgrowth of the distal humerus may result in bone exposure, requiring bone debridement and soft tissue closure. Some procedures need to be repeated until humeral growth ceases.²¹

Function may be improved by releasing syndactylized digits and web space contractures. Tendon transfers may improve opposition in selected cases. In severe cases, stump lengthening, through bone grafting or distraction osteogenesis, has been performed to facilitate prosthetic fitting. In rare cases in which the hand is severely deficient but a forearm component is present, a Krukenberg procedure has been recommended to provide prehension.²¹

KEY POINTS

- Evaluating a child's functional capacity is the most important step in developing a treatment plan.
- Prosthetic management is only infrequently accepted by children with congenital differences of the upper extremities.
- Some patients lose function after centralization of the wrist.
- Centralization of the wrist will probably be unsuccessful if preoperative stretching and splinting does not achieve passive correction of the deformity.
- Prolonged splinting may be required to maintain a centralized position.
- Patients with cleft hand usually have good function that should not be impaired by efforts to improve cosmesis.
- Intermetacarpal movement is critical for success in toe-to-hand transfer in symbrachydactyly patients.
- The entire extremity, including the chest wall, should always be examined.

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43

The Hypoplastic Thumb and Absent Thumb

Joseph Upton III • Christopher J. Coombs • Daniel J. Wilks



child with thumb hypoplasia may present many unique problems with both grasp and digital prehension. The anatomic abnormalities seen with small or absent thumbs span a continuum ranging from minimal muscle and size discrepancies to complete aplasia. A thumb should be considered hypoplastic when there is a deficiency of any one or all of the structures that contribute to the normal structure.

Deficiencies of the axial skeleton, joints, intrinsic or extrinsic myotendinous units, or the soft tissue envelope can occur as an isolated deformity, in unison, in conjunction with other problems in the hand, or as part of generalized syndromes. When considered as a single diagnostic entity, thumb hypoplasia is one of the most common congenital conditions treated by hand surgeons. In our registry of approximately 8200 hands treated at the Boston Children's Hospital, hypoplastic or absent first rays are listed as a primary or secondary diagnosis in 2488 of the hands.

The impact of a hypoplastic or an absent thumb on hand function is not only related to the degree of the deformity but also to the severity of additional problems in the ipsilateral extremity. Cognitive function and cortical representation of the thumb will also have a great bearing on the child's ability to use his or her thumb. However, patients with severe central neurologic deficits may not benefit from the most sophisticated and elegant reconstructions. Decisions made in the treatment of these children directly affect not only the child, but also the family, physicians, and other care providers, who should be consulted when appropriate.

EMBRYOLOGY

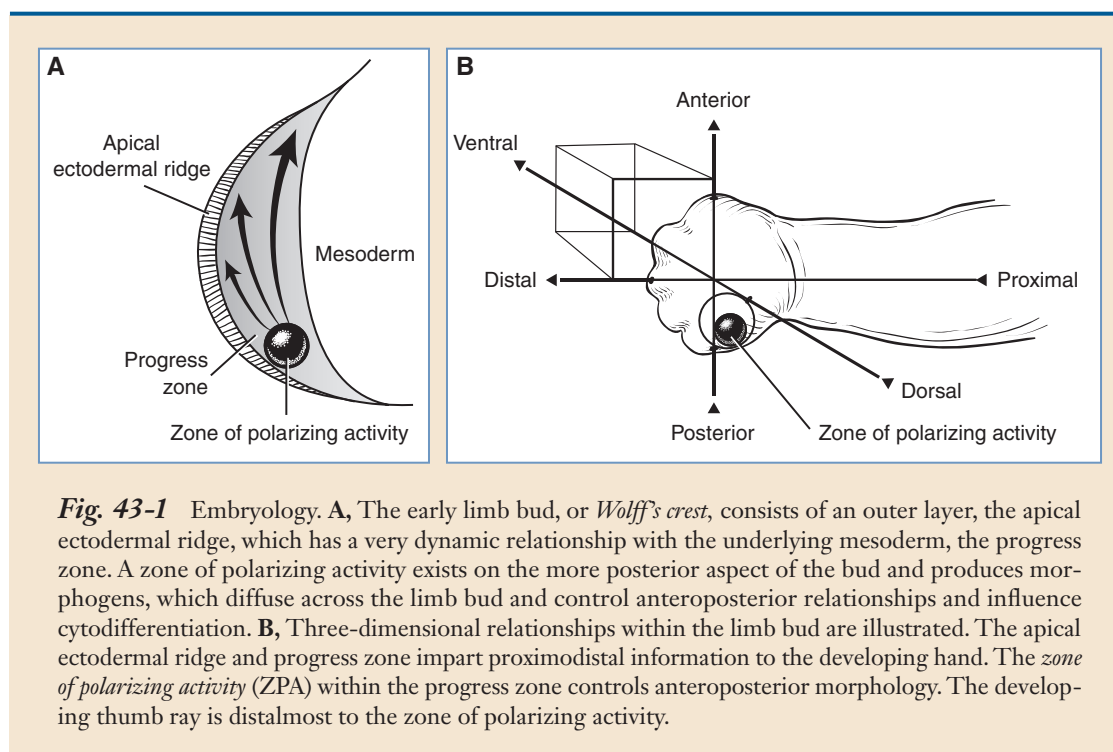
Since Saunders' classic experiments performed on chicken embryos over 60 years ago,¹ much has been discovered about the intricate interplay of genes, proteins, and transcription factors that consistently produce the recognizable and vastly complex human limb. Although many of the molecular mechanisms are yet to be fully elucidated, great advances in the understanding of limb development have recently occurred through the application of molecular biology, animal models, and clinical genetics.

Streeter² described vertebrate embryo development as occurring along a preprogrammed path of 23 steps (called *Carnegie stages*). By day 30 of embryogenesis (Carnegie stage 9), the upper limb bud (at this stage called *Wolff's crest*) appears as a swelling on the ventrolateral sides of the embryo. By day 56 (Carnegie stage 23), nutrient vessel penetration of the humerus and distal phalangeal ossification signifies the conclusion of embryogenesis. Formation of all structures within the upper limb is complete.

During the fetal period that follows, growth of the upper limb occurs differentially.

Development of the limb (Fig. 43-1) progresses along three axes: proximodistal, anteroposterior (or radioulnar), and dorsoventral (or dorsovolar).³ Differentiation and formation along each axis is coordinated by chemical cascades initiated by discrete signaling centers.⁴

Proximodistal growth is thought to be coordinated by the apical ectodermal ridge (AER). This is an area of thickened, specialized ectoderm that covers the underlying mesoderm, known as the *progress zone* (PZ). The AER caps the developing limb bud and primitive hand plate as it forms, until separation of the digits occurs. Saunders' experimental work showing that earlier removal of the AER produced more proximal limb truncation led to the progress zone hypothesis of proximodistal formation. The presence of an AER-secreted morphogen was suggested by



Wolpert,⁵ which through its action on the underlying PZ maintained the cells in an uncommitted state. As the limb enlarges, cells too proximal to be influenced by the diffusible morphogen then differentiate. Molecular studies have since confirmed Wolpert's assertion and identified fibroblast growth factor (FGF) to be the morphogen.⁶

However, selective genetic knockout of the *FGF* gene does not replicate the clinical effects of surgical ablation of the AER as observed by Saunders.¹ Rather, these embryos continue to develop distalmost features, yet have a reduction of proximal structures.^{7,8} This finding, taken with other studies, has led to a shift away from the progress zone hypothesis and development of new theories, such as the early specification (all components of the limb are present early and expand with growth) and the two-signal model (an additional signal, perhaps retinoic acid, is secreted proximally).⁹⁻¹¹

Digit number and digit-specific morphology is established by the anteroposterior axis, which is governed by a further area of specialized ectoderm, found in the axilla of the developing limb bud within the PZ; that is, the ZPA. Consistent with the mechanisms of *FGF*-derived proximodistal growth, anteroposterior patterning occurs as a result of a diffusion driven, activator/inhibitor instability model; in this situation governed by *Sonic Hedgehog* (*SHH*) expression from the ZPA.¹²

SHH manifests its action through *HOXA* and *HOXD* genes. Whereas *HOXA13* and *HOXD13* are found in all digits, *HOXD10* through *HOXD12* are expressed in the fingers only.¹³ *HOXD13* displays a relatively limited response to *SHH*.

SHH also acts to inhibit truncation of the *GLI3* transcription factor. An absence of *SHH*, as found within the anterior hand plate, is therefore associated with an abundance of the truncated form (*GLI3R*), which is a strong transcriptional repressor. A posterior to anterior *SHH* gradient is therefore generated along with a reciprocal anterior to posterior *GLI3R* gradient. Increased levels of *GLI3R* and reduced levels of *SHH* on the anterior aspect of the hand plate are thought to lead to thumb development through second messenger mechanisms.

Specific defects in genes regulating limb patterning are associated with recognized thumb anomalies. For example, mutation of *HOXA13* causes hand-foot-uterus syndrome, which manifests proximally based, hypoplastic thumbs,¹⁴ disruption of *TBX5* causes Holt-Oram syndrome,¹⁵ whereas *SALL1* and *SALL4* mutations are associated with Townes-Brocks and Duane-radial ray syndrome.¹³

The signaling center associated with the dorsoventral axis, located in the dorsal ectoderm, produces a protein called Wnt-7a. This induces production of transcription embryology and classification factor *Lmx1* which is required for dorsalization of the limb.¹⁶ Once more, an activator inhibitor loop is generated by an additional transcription factor, produced on the ventral aspect of the limb termed *engrailed-1* (*En-1*) which blocks Wnt-7a expression.¹⁷ Wnt-7a also promotes *SHH* in the ZPA and thus has a role in both proximodistal and anteroposterior growth.

The normal development and formation of the thumb can be affected either during embryogenesis (fertilization to 8 weeks) or during fetal growth (8 weeks to birth). Even though the majority of mechanisms by which congenital differences of thumb development (and upper limb) occur are not known, most congenital differences are felt to occur during embryogenesis. These may result from chromosomal abnormalities, inborn errors of metabolism, infections, or teratogens.¹⁸ Subsequent necrosis of cells vital to the correct formation of the upper limb or delays in programmed cell death may then result in abnormalities of development (failure of formation: for example, in the fetus of a woman who has taken thalidomide^{19,20}; failure of differentiation: for example, syndactyly²¹). Following formation of the limb, abnormal rates of growth during the fetal period may result in hypertrophy (overgrowth: for example, nerve territory oriented macrodactyly) or hypoplasias (undergrowth: for example, brachydactyly). Congenital amputations seen in the amniotic band sequence (Streeter's dysplasia) occur late within the fetal period and are classified by pediatric geneticists as deformations of a normal hand plate that occur in utero. In

this sequence the secondary deformations to normal structures occur with changes in the uterine environment. Chromosomal and other inborn errors are designated as malformations. A disruption sequence occurs when normal tissue is subjected to an infection, or by vascular, mechanical, or metabolic toxins such as the sedative thalidomide, which was responsible for hypoplastic limbs five decades ago, primarily in Europe.²² Approximately 15% to 20% of thumb anomalies have a genetic chromosomal cause, most of which should be identified or mapped within the next decade. No more than 30% of the anomalies have known infectious, metabolic, or other identified causes, and for the largest group the cause remains unknown. A comprehensive exposition of the factors currently thought to be important in limb development can be found in reviews by Zeller et al²³ and Al-Qattan and Kozin,²⁴ whereas Oberg¹³ specifically considered thumb development.

Because of these developments, reappraisal of Swanson's classification of limb anomalies²⁵ is a logical progression of a greater understanding of limb development. Classifications based on the effects of specific genetic or molecular defects, such as that suggested by Tonkin et al,²⁶ are likely to become both more specific and complex as our understanding of this carefully choreographed process progresses. However, the chromosomal aberrations do not provide clear-cut pathways for the determination of clinical decision-making.

FREQUENCY

The frequency of congenital differences is subject to variation because of the genetic composition of the specific population being reviewed as well as discrepancies in nomenclature and sampling.²⁷⁻³⁰ Congenital differences involving the upper limb have been reported to occur in 1:200 to 1:626 live births. Entin^{31,32} observed that major congenital differences involving the thumb occurred in 16% of his series of congenital upper limb abnormalities. In 1977 Flatt³³ reported the frequency of hypoplastic thumb within his Iowa practice of congenital anomalies of the upper limb to be 3.6%. Lamb and colleagues,^{34,35} in their patients with 117 radial longitudinal deficiencies, found that there was variable penetrance and expressivity of thumb hypoplasia. Thumb hypoplasia is also associated with other hand anomalies and generalized syndromes involving the hematologic, musculoskeletal, cardiovascular and genitourinary systems. These are listed in Box 43-1.³⁶ Problems with small or inadequate thumbs are some of the most common encountered in congenital hand surgery, including hypoplastic thumbs seen in cleft hands, preaxial duplications, the constriction ring syndrome, clasped thumbs, retroflexed thumbs, five-fingered hands, and other diagnoses.³⁷ Therefore consultation with a genetic specialist is strongly recommended, and referral to standard genetics textbooks is a must for any responsible hand surgeon.

CLASSIFICATION

The classification of hypoplastic thumbs is presently determined by the clinical appearance of the affected thumb. The anatomic characteristics of the first ray form the basis of our current classification system. Before classification and management of a child with a hypoplastic thumb, a careful assessment includes the following:

- Size
- Position
- Relation to other fingers in the hand
- Osseous components
- Joint integrity
- Intrinsic and extrinsic myotendinous units
- First web space depth and width
- Associated hand anomalies

Box 43-1 Thumb/Radial Hypoplasia/Aplasia Associations**Thumb (T)/Radial (R) Hypoplasia/Aplasia**

Frequently seen in:

Aase syndrome (T, R)
 Baller-Gerold syndrome (T, R)
 Congenital microgastria-limb reduction complex (T)
 Deletion 13q syndrome (T)
 EEC syndrome (T)
 Fanconi syndrome (T, R)
 Gómez-López-Hernández syndrome (T, R)
 Holt-Oram syndrome (T, R)
 Levy-Hollister syndrome (T, R)
 Miller syndrome (R)
 Nager syndrome (T)
 Oculo-auriculo-vertebral spectrum (T)
 Okihiro syndrome (R)
 Radial aplasia-thrombocytopenia syndrome (T, R)
 Roberts syndrome (T, R)
 Rothmund-Thomson syndrome (T)
 Townes syndrome (T)
 Ulnar-mammary syndrome (R)
 VACTERL Association (T, R)
 Yunis-Varón syndrome (T)

Occasionally seen in:

Brachmann-de Lange syndrome (T, R)
 Cat-eye syndrome (R)
 CHARGE syndrome (T)
 Fetal aminopterin/methotrexate syndrome (T)
 Fetal valproate syndrome (T, R)
 Fibrodysplasia ossificans progressiva syndrome (T)
 Fraser syndrome (T)
 Fryns syndrome (T)
 Laurin-Sandrow syndrome (R)
 Lenz microphthalmia syndrome (T)
 Miller syndrome (T)
 MURCS association (T)
 Nager syndrome (R)
 Popliteal pterygium syndrome (T)
 Seckel syndrome (R)
 Smith-Lemli-Opitz syndrome (R)
 Trisomy 13 syndrome (R)
 Trisomy 18 syndrome (T, R)

Metacarpal Hypoplasia

Frequently seen in:

CHILD syndrome (all)
 Coffin-Siris syndrome (all)
 Cohen syndrome (all)
 Deletion 18q syndrome (first)

Deletion 2q37 syndrome (all)
 Deletion 4q syndrome (first)
 Deletion 5p (cri du chat) syndrome (all)
 Diastrophic dysplasia (first)
 Duplication 10q syndrome (first)
 Duplication 9p syndrome (all)
 Dyggve-Melchior-Clausen syndrome (all)
 Grebe syndrome (all)
 Microdeletion 15q24 syndrome (first)
 Oto-palato-digital syndrome type II (all)
 Poland sequence (all)
 Short rib polydactyly, Majewski type (all)
 Tricho-rhino-phalangeal syndrome type I (all)

Occasionally seen in:

Brachmann-de Lange syndrome (first)
 Larsen syndrome (all)
 Multiple epiphyseal dysplasia (all)
 Robinow syndrome (all)
 Tibial aplasia-ectrodactyly syndrome (first)
 Triploidy syndrome and diploid/triploid mixoploidy syndrome (first)

Broad Thumb

Frequently seen in:

Aarskog syndrome
 Acromesomelic dysplasia
 Apert syndrome
 Carpenter syndrome
 Curry-Jones syndrome
 FG syndrome
 FGFR3-associated coronal synostosis syndrome
 Floating-Harbor syndrome
 Greig cephalopolysyndactyly syndrome
 Larsen syndrome
 Microdeletion 2q31.1 syndrome
 Oto-palato-digital syndrome, types I and II
 Pfeiffer syndrome
 Rubinstein-Taybi syndrome
 Saethre-Chotzen syndrome
 Simpson-Golabi-Behmel syndrome
 Weaver syndrome

Occasionally seen in:

Dubowitz syndrome
 Lenz microphthalmia syndrome
 Matthew-Wood syndrome
 Robinow syndrome
 Trisomy 13 syndrome

CHILD, Congenital hemidysplasia with ichthyosiform erythroderma and limb defects; *EEC*, ectrodactyly, ectodermal dysplasia, and clefting; *MURCS*, Müllerian duct hypoplasia/aplasia, renal agenesis, and cervicothoracic somite dysplasia; *VACTERL*, vertebral anomalies, anal atresia, cardiac defect, most often ventricular septal defect, tracheoesophageal fistula with esophageal atresia, renal abnormalities, limb abnormalities, most often radial dysplasia.

Complete evaluation of the osteoarticular column will require radiologic investigation. Even though primary ossification centers of the phalanges and metacarpal of the thumb appear in the second to fourth fetal months, abnormalities of the skeleton of the thumb, for example, triangular bones, may not be seen radiographically until well into the first or second year of life. In addition, secondary ossification centers within the epiphyses of the thumb normally appear between 13 months and 4 years of age.³⁸ The appearance of both primary and secondary ossification centers in a hypoplastic thumb is often delayed in proportion to the degree of hypoplasia. A delayed appearance should be expected in most thumb differences beyond type I³⁹ (Fig. 43-2).

Because there is a spectrum of thumb hypoplasia, accurate classification of the abnormality is required so that an appropriate management plan can be initiated. The current classification system that was proposed by Müller⁴⁰ and Blauth and Schneider-Sickert^{41,42} was expanded by Buck-Gramcko⁴³ in 1981 and defined five types of thumb hypoplasia and aplasia. This classification recognizes the progressive degree of hypoplasia from a slight size discrepancy, with all normal structures present, to total aplasia of the thumb. In 1995 Manske et al⁴⁴ proposed their modification of this system, dividing type III hypoplastic thumbs into IIIA and IIIB. This distinction is made by the presence (IIIA) or absence (IIIB) of a stable carpometacarpal joint (Box 43-2) and is very important in terms of management. These five types of thumb hypoplasia are

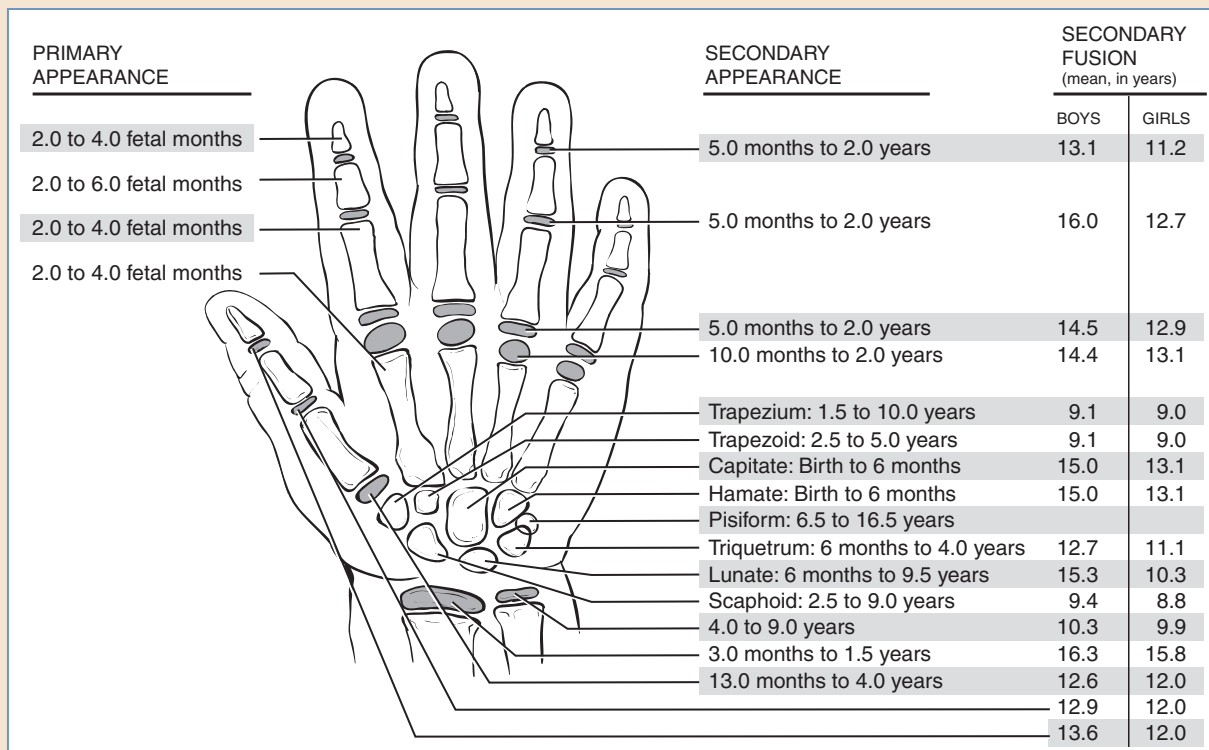


Fig. 43-2 Ossification times. Normal primary and secondary ossification centers of the hand are diagrammed with their times of radiographic appearance. At birth, primary ossification is present for the thumb metacarpal and phalanges. The secondary centers for the phalanges appear much later. Within the hypoplastic thumb or hand, the appearance of all ossification centers may be significantly delayed. There is also considerable variation between sexes and different populations.

Box 43-2 Classification of Thumb Hypoplasia**Type I: Mild Hypoplasia**

Minimal skeletal shortening
 Intrinsic muscle intact
 Extrinsic muscles intact
 Tendons intact

Type II: Moderate Hypoplasia

Moderate skeletal shortening
 Intrinsic muscle hypoplasia (median innervated)
 Small first web space
 Adducted thumb position
 MP joint instability

Type III: Severe Hypoplasia

Severe skeletal shortening
 Carpal hypoplasia
 Intrinsic aplasia (median) and hypoplasia (ulnar)
 Extrinsic tendon abnormalities
 Small first web space
 Severe MP joint instability

Type IIIA

Metacarpal very hypoplastic
 Carpals present
 CMC joint intact

Type IIIB

Proximal metacarpal aplasia
 Trapezium absent
 CMC joint absent

Type IV: Pouce Flottant (Floating Thumb)

Metacarpal absent; phalanges rudimentary
 Skin and neurovascular connection
 Intrinsic, extrinsic tendons absent

Type V: Aplasia

All structures absent

Type VI: Cleft Hand (Ectrodactyly)

Metacarpal, phalanges present
 First web syndactyly common; all degrees of syndactyly
 Deficient or absent adductor pollicis
 Thenar intrinsic (median innervated) present
 Skeleton hypoplastic
 Occasional thumb polydactyly

Type VII: Symbrachydactyly (Atypical Cleft Hand)

Central three rays most deficient; all degrees of hypoplasia to aplasia of index; long and ring rays; digits are often hypoplastic nubbins
 U-shaped cleft
 Flexor pollicis abnormal, often with intrinsic origin
 Ulnar-innervated intrinsic muscles to thumb are often absent
 Median-innervated intrinsics are usually present
 IP joint is stiff; MP joint is present and occasionally unstable
 Extrinsic flexor and extensor muscles are often weak
 Fifth ray is the best in the hand
 Hypothenar muscles intact

Type VIII: Thumb in Constriction Ring Syndrome (CRS)

Transverse amputation at level of:
 IP joint
 Proximal phalanx
 Metacarpal
 Anatomy proximal to amputation is normal
 Constriction rings (annular bands) at any level

Type IX: Thumb Hypoplasia in Polydactyly

Skeletal level and degree of complexity increases from distal to proximal
 Osteoarticular column is hypoplastic in biphalangal thumbs, higher than type IV (Wassel classification)
 Nail bed is smaller than normal in both partners
 Extrinsic tendons are shared
 Intrinsic muscles are present with abnormal insertions

Type X: Thumb Hypoplasia in the Five-Fingered Hand

Normal ray with metacarpal and three phalanges
 Absent thenar (median-innervated) intrinsics
 Normal digital intrinsics and extrinsics

Type XI: Short Skeletal Ray

Isolated or combined deficiency of metacarpal, proximal, or distal phalanges
 Collateral ligaments intact
 Triangular bones ("delta" phalanges) with longitudinal bracketed epiphysis are common
 ACS syndromes: Apert, Pfeiffer, Rubinstein-Taybi, and others
 Intrinsic muscles are intact, with abnormal insertions

commonly associated with radial (preaxial) dysplasia, and most congenital hand surgeons consider hypoplasia of the thumb with a normal radius as part of this spectrum. We have found it quite useful to correlate the degree of hypoplasia with the median- and ulnar-innervated thenar musculature (Fig. 43-3).

Hypoplasia of the thumb is also associated with many other congenital differences of the hand. Because the anatomic makeup of the thumb associated with these congenital conditions does not always allow easy categorization under the current classification system, we recognize six

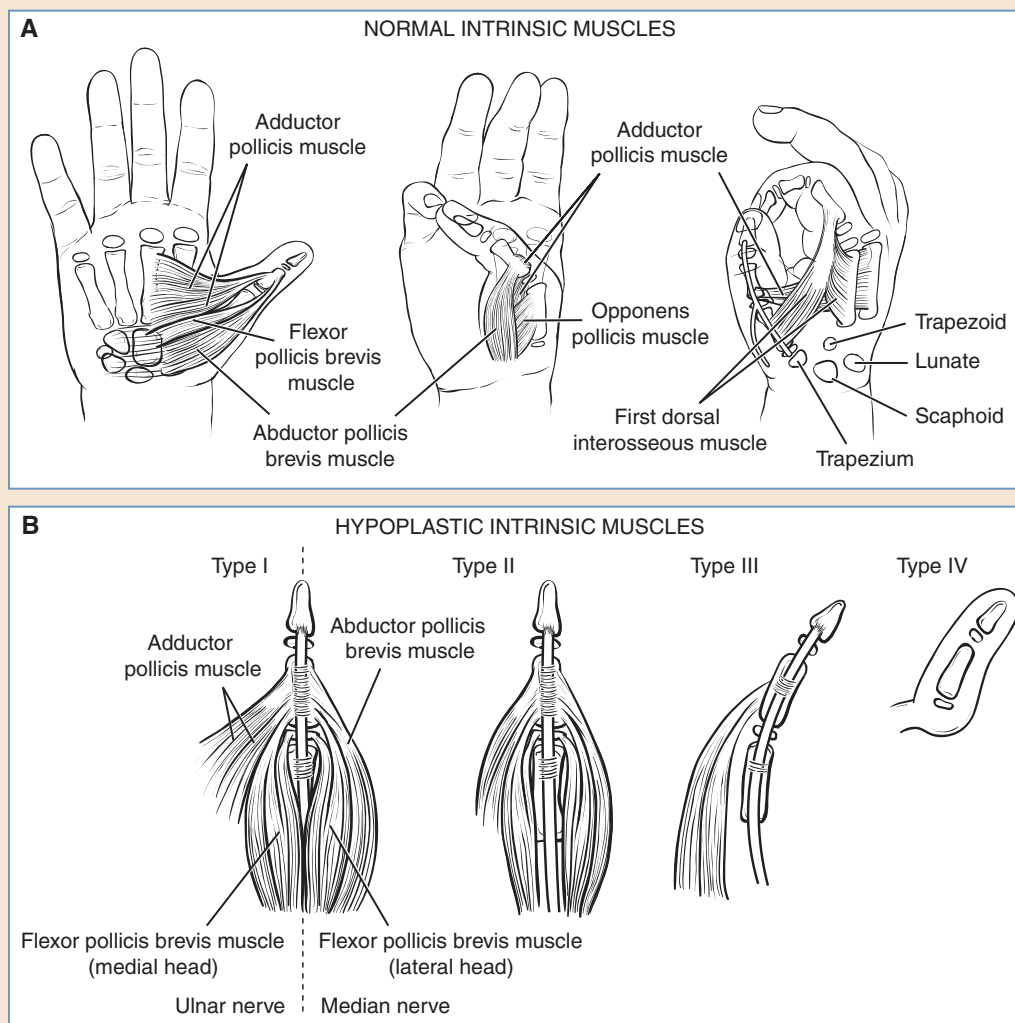


Fig. 43-3 Thumb hypoplasia. **A**, The normal median-innervated intrinsic muscles (abductor pollicis brevis, flexor pollicis brevis/lateral head, and opponens pollicis) and ulnar-innervated intrinsic muscles (adductor pollicis, first dorsal interosseous, and flexor pollicis brevis/medial head) to a right thumb are illustrated. **B**, Within the spectrum of thumb deficiencies seen with radial dysplasias, all intrinsic muscles are present in type I thumbs and are severely hypoplastic or absent in type III thumbs. Only the ulnar-innervated adductor pollicis and the flexor pollicis brevis medial head are present and functional in hypoplastic type III thumbs. Rudimentary muscles may be present in type IV floating thumbs. The first dorsal interosseous muscle (also called the *abductor indicis* muscle), with its second metacarpal origin, is found only when the index digit is present.

other types of hypoplastic thumbs. These are associated with cleft hand, symbrachydactyly, constriction ring syndrome, radial polydactyly, five-fingered hand, and generalized syndromes with short, commonly angulated, osteoarticular columns, such as the acrocephalosyndactylies (see Box 41-2). Other authors consider most of these additional categories as either type I or II thumbs.

TIMING

As researchers and clinicians struggle with the possibilities of fetal surgery, many surgeons wonder whether they should be performing reconstructive procedures of the congenitally different hand as early as possible. Because the thumb accounts for 50% of hand function, early reconstruction of a hypoplastic or an absent first ray is certainly attractive so that the infant can develop with optimal cortical representation and use of the thumb. This ideal needs to be tempered with the knowledge that a congenital hand difference is in itself not a life-threatening condition (but may be associated with one), and the surgeon can use time to allow the affected part to grow, to observe its development, and to assess functional needs. A stable osteoarticular column of adequate length, mobile joints, growth potential, and an adequate, scar-free first web are the necessary requirements for performing surgery on a hypoplastic or absent thumb. If these requirements can be met, then with the use of magnification and adequate microsurgical skills, precise yet demanding surgical procedures can be performed on very young patients.

Many other factors need to be considered. Several arguments that are forwarded by the proponents of early surgery include the following^{37,45}:

- Anatomic: Release of tethered myotendinous units and joint contractures will help allow unrestricted growth, and anatomic adaptation of the reconstructed thumb will occur secondary to growth and functional use.
- Cognitive: Early surgery will allow the development of the child with a reconstructed thumb before thumb corticalization, which occurs at about 18 months of age.
- Psychological: Potential alleviation of anxiety in the parents and therefore the child.

These advantages of early surgery need to be weighed against those for delayed procedures, which include the following:

- The affected thumb is larger in size so that potential problems with osteotomies and fixation, joint reconstruction, and vascularity are reduced.
- A better assessment of the functional needs of the child can be performed.
- The child is potentially a more cooperative patient.

Reconstruction of the thumb should be completed by the time the child starts school so that maximal functional benefits can be achieved and the potential for ridicule by peers is minimized. In the absence of other organ system complications, we try to perform reconstructive procedures on hypoplastic or absent thumbs when the child is between 10 and 18 months of age. Pollicization at 1 year is often preceded by centralization or radialization of the hand and wrist between 5 and 8 months of age. In children with radial deficiencies and thumb aplasia of one upper limb and a less severe thumb hypoplasia on the opposite limb, surgeons should treat the stronger side, with the hypoplastic thumb, before attempting pollicization on the more deficient side. Because this hypoplastic thumb will be the child's best thumb, it is important to strengthen and improve it as soon as possible. However, parents often prefer to have the pollicization performed first and to have nothing done on the more complete side. Occasionally, simultaneous bilateral surgery is appropriate, for example, in a child with a type II thumb on one side and a contralateral type IV or V thumb.

The timing for correction of type IIIB deformities may be problematic. Although we believe pollicization is the procedure of choice, some parents and families simply will not allow it. We agree with other surgeons that the alternatives involve difficult reconstructions, often including two or more stages, and are wisely deferred until the child is 4 to 5 years of age, a time when the hand is larger and the patient may be more cooperative with the postoperative program.⁴⁶⁻⁴⁹

The determination of the optimal time to reconstruct a hypoplastic or absent thumb must be individualized in each patient. As these children may have other congenital anomalies associated with their thumb abnormality, it is important to assess these patients early in life so that a coordinated management plan can be formulated and instituted. Perhaps the most important variable is the confidence, surgical skill and experience of the surgeon and his team.

TYPE I THUMB HYPOPLASIA: MILD HYPOPLASIA

Anatomy

Type I represents the mildest form of hypoplasia within this group of congenital thumb differences. The thumb is slightly smaller but is normally configured (Fig. 43-4). The phalanges and metacarpal of the thumb can be slightly thinner than usual. The trapezium and scaphoid are present and the distal radius and styloid are not affected. The interphalangeal (IP), metacarpophalangeal (MP) and carpometacarpal (CMC) joints of the thumb are stable and exhibit normal passive and active motion. The intrinsic and extrinsic myotendinous units are present. The

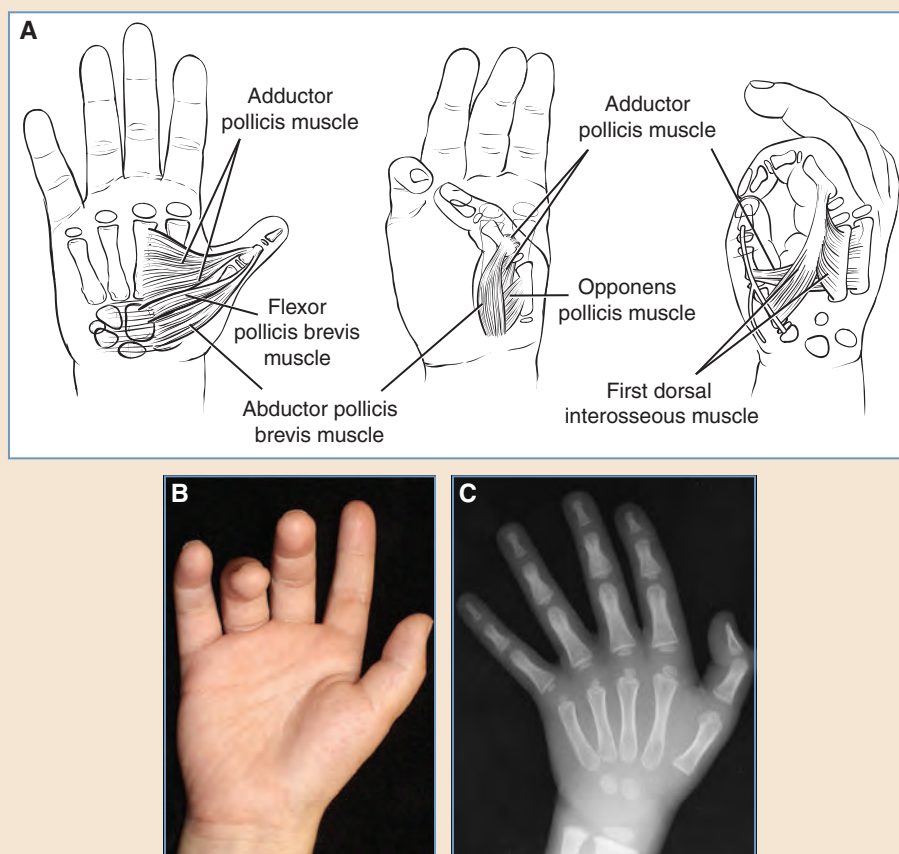


Fig. 43-4 Type I thumb hypoplasia—mild. **A**, The skeletal ray is well segmented and may be short. All intrinsic muscles and extrinsic tendons are intact. First web space narrowing is minimal to moderate. **B** and **C**, Clinical and radiographic appearance of a child who did not require any surgical correction. Although his thenar muscles are weak, there are no significant functional problems.

median-innervated intrinsic muscles (flexor pollicis brevis [FPB]–lateral head, opponens pollicis [OP], and abductor pollicis brevis [AbPB]) can occasionally be mildly hypoplastic. The first web space may be slightly narrowed.

Management

These children are not usually functionally impaired. In fact, many children with type I thumb hypoplasia do not recognize anything abnormal about their hands. These thumbs are commonly found in patients who have a more severe radial dysplasia involving the contralateral upper limb. Children with type I hypoplasia have very little or no difficulty with key, pulp, or nail pinch, opposition, and grasping activities. Because functional problems are rare, surgical correction is not needed. Very occasionally a child with a type I hypoplastic thumb will require a release of a mildly contracted web (see Management under Type II Thumb Hypoplasia: Moderate Hypoplasia in this chapter). Any other procedures need to be precisely determined and planned to meet the functional needs of the patient.

TYPE II THUMB HYPOPLASIA: MODERATE HYPOPLASIA

Anatomy

Children with type II thumb hypoplasia have a moderate diminution in the size of the affected thumb (Fig. 43-5). The osteoarticular column is narrowed and short. The trapezium and scaphoid are present but may be small and may result in slight radial deviation of the hand. The distal radius has a normal configuration but may be short on occasion. Characteristic of type II thumbs is instability of the MP joint. This particularly effects the ulnar side of the joint with severe laxity of the ulnar collateral ligament (UCL). The IP and CMC joints are stable, but both may have some mild limitation of motion. The median-innervated intrinsic muscles are hypoplastic in contrast to the ulnar-innervated intrinsics, which are usually normal (see Figs. 43-4 and 43-5). This asymmetrical hypoplasia of the intrinsic musculature causes a dynamic imbalance on either side of the thumb. The stronger ulnar-innervated intrinsics, and in particular the adductor pollicis (AddP), adducts the thumb and contributes to a narrowing of the first web space. The degree of this deficiency is usually a reflection of the degree of hypoplasia of both the skeleton and the median-innervated intrinsics.

The pollex abductus deformity may be present and represents anomalous connections between the flexor and extensor tendons along the radial border of the thumb (Fig. 43-6). Uncommonly, a proportion of these patients will have absent^{50,51} or anomalous insertions^{47,52-55} of the flexor pollicis longus (FPL). The FPL or EPL tendon can also be in an aberrant position lying along the radial border of the proximal phalanx. When present, these tendons pass superficial to the hypoplastic thenar muscles, to which they are often connected. Distally the anomalous tendon usually inserts by two slips, one large slip into the volar-radial base of the distal phalanx and the other into the extensor pollicis longus tendon. The malpositioned FPL and/or EPL tendon functions as an abductor of the MP joint. Continued pinching activities associated with the anomalous FPL and the instability of the ulnar collateral ligament will result in radial deviation of the phalangeal portion of the thumb with concomitant adduction of the first metacarpal.

Other tendon anomalies seen with type II hypoplasia include eccentric insertions of extrinsic flexor and extensor tendons resulting in IP joint deviation, abnormal extensors,^{55,56} aplasia of the thenar muscles,⁵⁷ and occasionally anomalous muscles.^{54,58} Although the degree of hypoplasia seen with duplications, triphalangeal thumbs, cleft hand, symbrachydactyly, five-fingered hands, and so forth fall into the type II category, we have separated this large group of patients into individual categories for ease of anatomic analysis and consideration of treatment options.

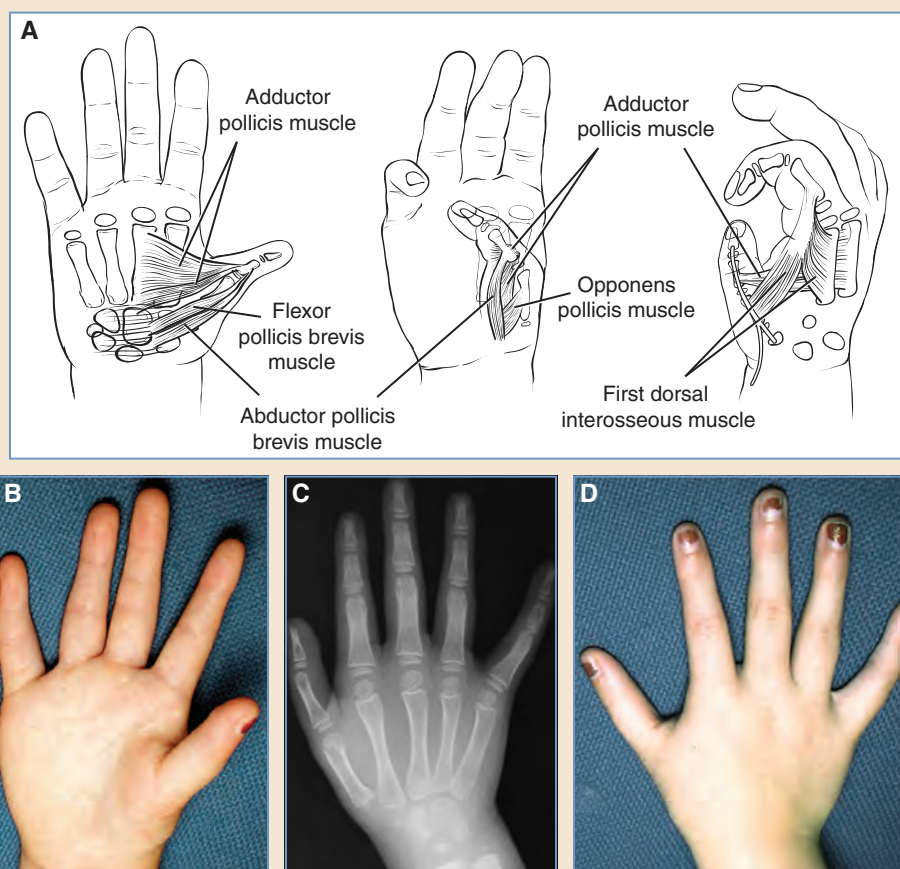


Fig. 43-5 Type II thumb hypoplasia—moderate. **A**, There is minimal to moderate shortening of the skeletal ray. All bones are present and ligaments at the MP joint may be lax. The ulnar-innervated intrinsic muscles, AddP, and first DI (second metacarpal origin) are strong, and the median-innervated intrinsic muscles are very weak. Anomalous anatomy is common. Adduction contractures from fibrous structures and MP joint instability accompany a deficient first web space. **B-D**, Clinical appearance and radiograph of a child with type II hypoplasia of the right hand. The thenar intrinsic muscles are hypoplastic, the first web space is tight, and the thumb is much narrower than normal. Key pinch is weak, and the patient cannot hold heavy objects between the thumb and digits. In addition, the ulnar collateral ligament is weak at the MP joint.

Management

The problems that need to be addressed in children with type II hypoplasia are (1) narrowed first web, (2) instability of the MP joint, (3) poor palmar abduction (opposition), and, if present, (4) a lack of flexion of the IP joint. These problems can be managed surgically in one or two stages. Flatt³³ preferred to stabilize the MP joint and release the first web in an initial procedure, then restore opposition in a second procedure, whereas others prefer to reconstruct type II thumbs in one stage.³⁷ It is particularly important to avoid damage to the growth plates in any of the tubular bones of the thumb as any degree of growth arrest early in development will adversely affect the outcome of any reconstruction.

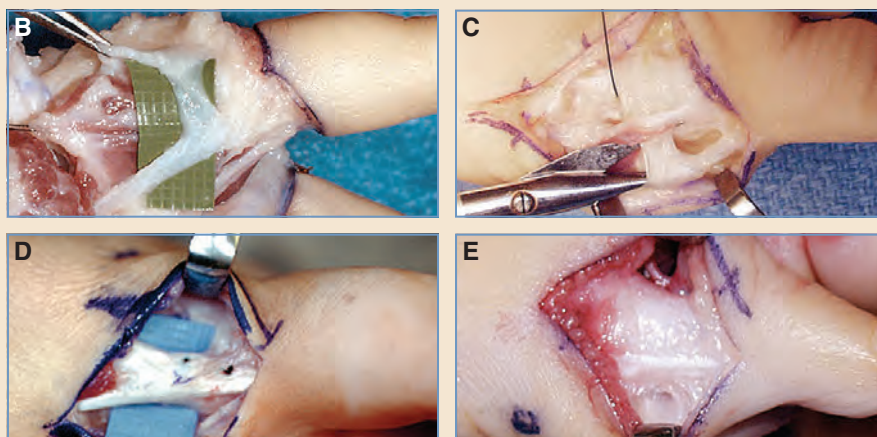
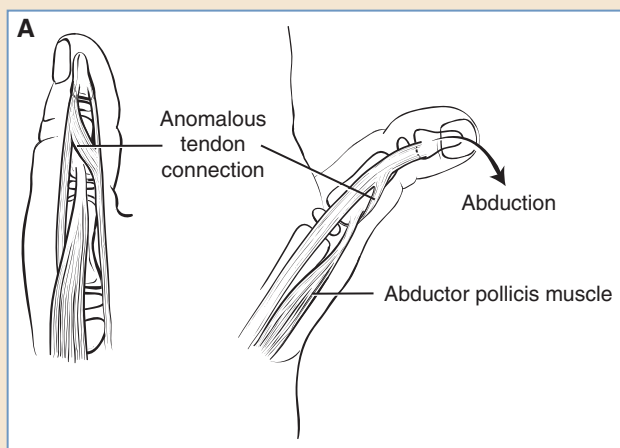


Fig. 43-6 Pollex abductus. **A**, In severe type II and many type III thumbs, there may be very little, if any, IP joint flexion, and there may be a radial deviation of the thumb at the MP joint because of the abduction force created by the combined action of the extrinsic flexor and extensor tendons joined by intertendinous connections, the pollex abductus. **B-E**, Within types II and III thumb hypoplasias (and proximal thumb duplications), a very wide spectrum of intratendinous connections may vary between a wide, loose, almost areolar band (*seen at top*) to a complete coalition of the flexor and extensor into a single tendon. With excursion from either the flexor or extensor side, the resultant movement in all these thumbs is abduction of the MP joint.

Narrowed First Web

The creation of an adequate first web space is of critical importance for the correction of any thumb anomaly and represents the most functional single correction in this group of patients. Normal thumb functions of key pinch, pulp and nail pinch, opposition, and thumb grasping maneuvers cannot be accomplished unless a good web space has been created. Many options for coverage are available and include various types of Z-plasty procedures, local flaps, local flaps with a full-thickness skin graft, tissue expansion, reverse-flow forearm island flaps, distant pedicle flaps, and free tissue transfer. Each of these procedures will provide different amounts and types

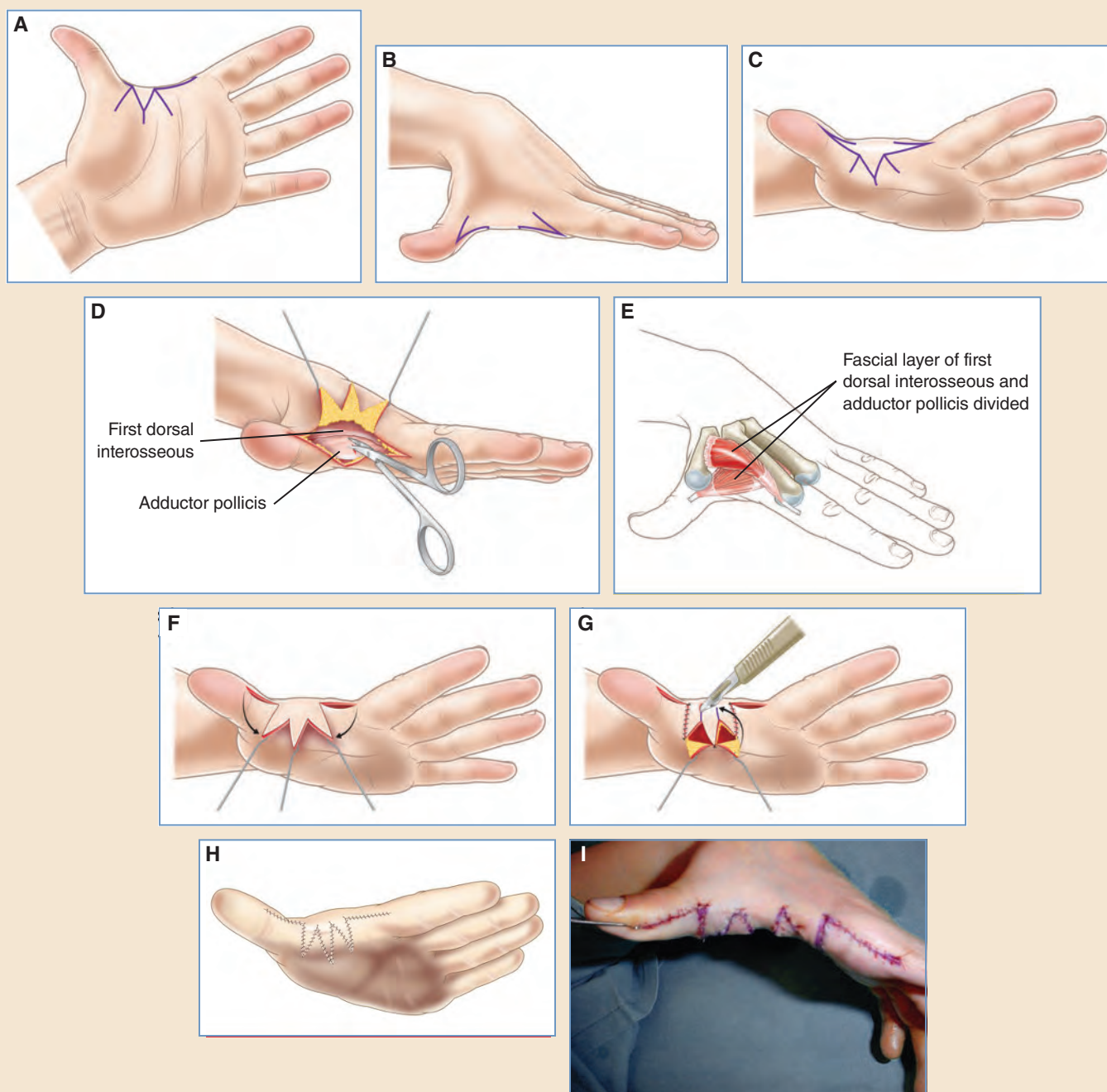


Fig. 43-7 First web space release—manta ray flap. **A-C**, The skin markings for the manta ray flap. **D** and **E**, The dorsal and volar fascia of adductor pollicis and the first dorsal interosseus muscles are released after the dorsal flap with the thumb and index extension have been raised. **F**, The dorsal flap is transposed to the volar surface and the markings of the volar flaps confirmed. **G**, Y-V advancement of the three limbs of the dorsal flap to widen the palmar web. **H**, Y-V dorsal advancement of the two volar flaps to release the dorsal web. **I**, Postoperative appearance; note the absence of palmar skin on the dorsum. This technique provides the best contour of all available methods of first web release.

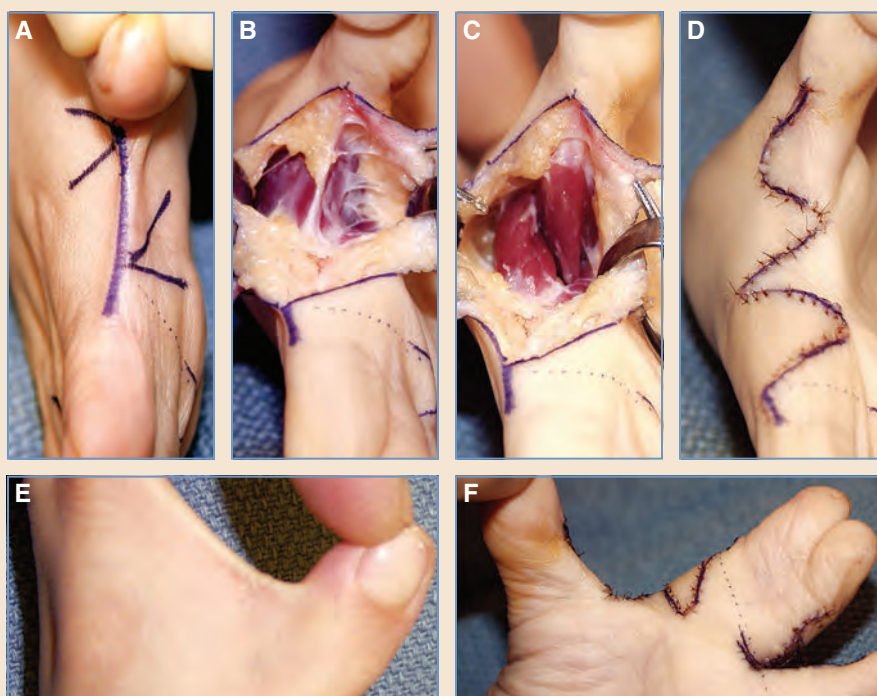


Fig. 43-8 First web space release—four-flap Z-plasty. **A**, The skin markings have been made for a four-flap Z-plasty, including two 90-degree incisions perpendicular to the tight web between the thumb and index finger. Each is then bisected at a 45-degree angle. **B**, After the four separate flaps have been mobilized, reflected dense septal bands may be identified between the skin and muscle fascia, and larger fascial bands between the intrinsic muscle groups. **C**, Incision and excision of these bands may require dissection to the CMC joint level. **D**, Trimming and minor flap adjustments are always needed with the proper inset of the more mobile dorsal flaps with the more rigid palmar flaps. A skin redundancy on the radial side of the index finger has been eliminated with a straight extension. **E**, Dorsal view of the preoperative appearance. **F**, The postoperative contour is shown from the palmar surface.

of tissue for coverage and the surgeon is required to determine which procedure is appropriate for each patient. In children with type II hypoplastic thumbs, local procedures, particularly the four-flap Z-plasty or the manta ray flap,⁵⁹ are usually sufficient to provide adequate coverage of the web following soft tissue release (Figs. 43-7 and 43-8). In our experience the manta ray flap provides a greater percentage release of the first web when compared to the four flap Z-plasty.⁵⁹

Web Release

Despite the type of flap repair that the treating surgeon has chosen for the resurfacing of the first web space, the release of the web deep to the skin is the same. The incisions required for the particular procedure chosen are marked and incised. The flaps are elevated with subcutaneous fat and with preservation of large dorsal veins and sensory nerves. The spreading dissection progresses directly to the fascia overlying the first dorsal interosseous muscle. With skin flaps reflected, the tight fascia enveloping the first dorsal interosseous muscle and a second fascial band between it and the adductor pollicis muscle are incised parallel to the first metacarpal toward the

CMC joint. This release usually produces a dramatic improvement of the web contracture. The radial digital nerve and artery to the index finger need to be protected. A similar layer of fascia on the volar surface of the adductor pollicis is released if present. Within the depths of the web, the radial artery may be located passing from dorsal to volar between the two heads of the first dorsal interosseous muscle and should be preserved. Injury to paired venae comitantes accompanying these arteries may result in unnecessary bleeding. The fascial release is then assessed with abduction of the index finger and thumb. Skin and fascial release alone are sufficient to provide an adequate release on type II thumbs.

Occasionally, release of other structures is required. The tightest muscle is usually the first dorsal interosseous muscle (first DI), which can be released and recessed from distal to proximal along its thumb metacarpal origin. With the index finger and thumb held in abduction, the adductor pollicis (transverse head) can be visualized passing directly across the web to its insertion on the ulnar side of the thumb. Recession of this muscle can be accomplished either by step lengthening of its adductor aponeurosis or by direct myotomy of the transverse head. Our preference is to release the transverse head from the third metacarpal, but we refrain from incising this muscle unless absolutely necessary. Occasionally, anomalous muscles such as the musculus lumbricalis pollicis are found and should be excised.⁵⁸ The CMC joint in type II thumbs is rarely a site of joint limitation, but if an established contracture exists, as is occasionally seen in a late-presenting teen or adult patient, release of the ulnar ligaments should be performed. Occasionally, a CMC release will dramatically improve thumb circumduction and will unmask a minimal to moderate instability of the MP joint.

First Web Skin Cover Options

In a patient with type II hypoplasia, the deficiency of skin within the first web is mild. Because of this most patients can be managed with the use of a Z-plasty. Two-, four-, and five-flap Z-plasties have all been advocated for skin coverage of the first web following its release.⁶⁰⁻⁶² The principle in each type of Z-plasty technique is the same in that they all lengthen the contractual diagonal of the Z-plasty by transposing tissue perpendicular to it. Other options for first web space coverage include the manta ray flap, a transposition flap from the dorsal and radial side of the index finger, a transposition flap from the dorsum of the thumb with skin graft of the secondary defect, sliding flap from the ulnar side of the index,⁵⁰ or dorsal rotation/sliding flaps with or without skin grafts for the secondary defects.^{63,64} The use of tissue expansion⁶⁵ (see Fig. 43-13) and reverse-flow forearm flaps for coverage of the contracted first web in congenital hand anomalies has been described.^{66,67} These latter procedures are applicable to more severe degrees of thumb hypoplasia and will be discussed later.

Preferred Method

We prefer to use the manta ray flap, which produces the most aesthetic and contoured web (see Fig. 43-7) without volar skin being present on the dorsal aspect of the first web—the socially presented side of the hand. The manta ray flap is marked as shown in Fig. 43-7. The central V of the flap is marked at the midpoint of the web. The ulnar thumb and radial index flaps are then marked. The width of the base of these is determined by a pinch test to ensure closure. The two digital and midweb flaps are raised together and release of the underlying structures performed. The central dorsal V flap is advanced through the web to its position of maximal advancement on the palmar web skin. This point is marked, then the palmar skin incised and the dorsal V flap is advanced in a Y-V fashion and sutured into position. The ulnar thumb flap is transposed into the palm with its maximal point of advancement marked and similarly incised and inset. The same procedure is done for the radial index flap. The donor sites on the thumb and index are then closed.

Following transposition and advancement of these three flaps, two palmar flaps are created and then these are advanced dorsally in a Y-V fashion to obtain dorsal release. Care needs to be taken to ensure appropriate placement of these incisions so the width of the base of the central dorsal V flap is not compromised. After full release of the web, the flaps are transposed and the wounds closed with 5-0 or 6-0 mild chromic catgut, depending on the patient's age.

Other methods of first web space augmentation will be illustrated with the more severe forms of thumb hypoplasia.

Metacarpophalangeal Joint Instability

Type II hypoplasia is characterized by mild-to-moderate instability of the MP joint, especially of the ulnar collateral ligament (UCL), which needs to be stabilized to prevent radial collapse of the thumb at the MP joint during pinch. The paired radial collateral ligament (RCL) on the opposite side of the joint may also be lax but rarely causes functional sequelae. In very unstable joints very few if any fibers of a normal collateral ligament are present and the existing structure has the gross appearance of an attenuated joint capsule.

Options

The options available to reconstruct the UCL are as follows:

- Reefing of the lax ligament and ulnar joint capsule
- Proximal advancement of the UCL on the metacarpal (if there is not a distal first metacarpal growth plate)
- Tendon graft
- Tendon transfer using the ends of the transferred tendon as a graft
- MP joint fusion

The major proviso of all of the collateral ligament reconstructions is that the growth plate at the base of the proximal phalanx and condyle of the metacarpal head must not be damaged. Frequently the first metacarpal of the thumb has a double epiphysis, which should be preserved to facilitate potential growth of the deficient thumb. Metacarpophalangeal joint fusion is used only in patients near the end of their growth.

Preferred Treatment

The stabilization procedure chosen for the UCL in patients with type II hypoplasia depends on the patient's age. In children less than 3 years of age, reefing of the ulnar side joint structures is preferred with K-wire stabilization of the MP joint. Proximal advancement and plication of the UCL on the metacarpal is done in those without a distal metacarpal growth plate. A free tendon graft, either sutured into the periosteum or anchored intraosseously, is used in virtually all other patients. This procedure is usually combined with an abductor digiti quinti muscle transfer for restoration of palmar abduction or opposition^{68,69} (Fig. 43-9). Occasionally, the flexor digitorum superficialis (FDS) from the ring finger is used for this purpose, and when transferred, the UCL is reconstructed with the terminal end of this tendon as part of the transfer (Fig. 43-10). Joint fusion is reserved as a late procedure in patients who are virtually full grown, have had failed soft tissue reconstructive procedures, have an asymmetrical metacarpal head with joint deviation, and the very occasional older patient with degenerative MP joint changes.

Chondrodesis of the MP joint tends to be an unpredictable procedure.⁷⁰ Because total active and passive range of motion is decreased in these patients, preservation of joint motion is always preferred. In patients who have significant radial deviation of the MP joint, the flexor pollicis longus tendon may well be displaced radially, and if so, it will need to be repositioned or reconstructed, since this can lead to recurrence of the deformity.^{53,54} The symmetrical head is flattened on the radial side and is common in these children with the pollex abductus anomaly.

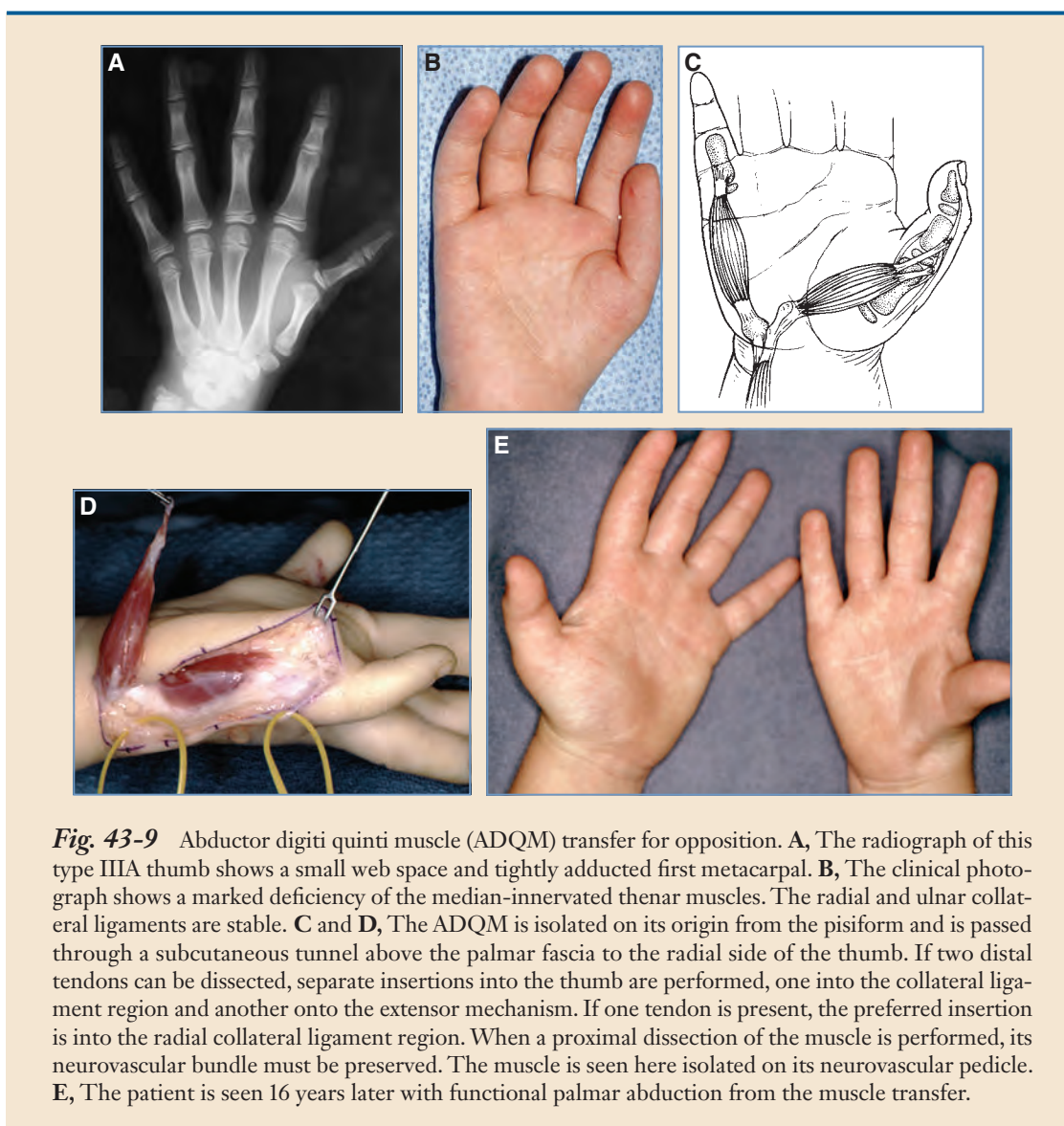


Fig. 43-9 Abductor digiti quinti muscle (ADQM) transfer for opposition. **A**, The radiograph of this type IIIA thumb shows a small web space and tightly adducted first metacarpal. **B**, The clinical photograph shows a marked deficiency of the median-innervated thenar muscles. The radial and ulnar collateral ligaments are stable. **C** and **D**, The ADQM is isolated on its origin from the pisiform and is passed through a subcutaneous tunnel above the palmar fascia to the radial side of the thumb. If two distal tendons can be dissected, separate insertions into the thumb are performed, one into the collateral ligament region and another onto the extensor mechanism. If one tendon is present, the preferred insertion is into the radial collateral ligament region. When a proximal dissection of the muscle is performed, its neurovascular bundle must be preserved. The muscle is seen here isolated on its neurovascular pedicle. **E**, The patient is seen 16 years later with functional palmar abduction from the muscle transfer.

Ulnar Collateral Ligament Stabilization

Following release of the first web, the ulnar side of the first MP joint is exposed. The dorsal sensory branches of the radial nerve to the thumb are retracted radially. Reefing of the attenuated ulnar collateral ligament and joint capsule can be addressed by either plication or by incision and closure of the lax structures with a “pants over vest” repair after dorsal-to-volar arthrotomy of the MP joint. The extensor mechanism with its attenuated shroud fibers is usually displaced radially and needs to be identified and mobilized from the dorsal joint capsule. The joint is held in full adduction and slight flexion with a single 0.028-inch K-wire. The ligament is then repaired with 4-0 or 5-0 nonabsorbable sutures. It is also important to tighten the attenuated dorsal capsule on the radial side of the joint. If not reefed snugly, relapse of the radial deviation of the thumb and subluxation of the proximal phalanx may occur.



Fig. 43-10 Thumb MP joint instability. **A**, The total absence of median-innervated thenar muscles and an unstable MP joint are characteristic of a type IIIA hypoplastic thumb. **B** and **C**, A well-segmented but flail joint is present. **D**, The superficial flexor tendon to the ring finger has been looped around the flexor carpi ulnaris and will be passed proximal to Guyon's canal toward the thumb. **E**, The transfer is secured tightly and both free ends are used to reconstruct the absent collateral ligaments. The *inset* shows the upper (cord) and lower (volar accessory) portions of the new radial collateral ligament. **F**, Three months postoperatively, the transfer is strong and the MP joint stable on both ulnar and radial sides.

Free Tendon Graft

Reconstruction of the UCL of the thumb MP joint with a free tendon graft in a child requires meticulous attention to detail so that the growth plates are not injured. Intraoperative radiologic assessment is essential to accurately place the tendon graft. The tendon graft is harvested from the ipsilateral palmaris longus; when this is absent, the choices are (1) contralateral palmaris longus, (2) plantaris, or (3) a toe extensor from one of the lateral three toes. The ulnar side of the MP joint is exposed and radiographs are taken of the thumb ray to determine the site of the metacarpal and proximal phalangeal epiphyseal plates. In a young child the tendon graft is sutured to the periosteum and is not passed through bone. This will minimize the risk of damage to the growth plates and prevent fracture of these slender bones. The tendon graft is first sutured to the proximal phalanx, away from its growth plate on its dorsoulnar aspect (at a point to mimic the insertion of the true collateral ligament). Next, the tendon graft is fixed to the volar-ulnar aspect

of the base of the proximal phalanx, again away from the growth plate (at a point to mimic the insertion of the volar accessory collateral ligament). Both ends are then brought proximally and sutured together to the dorsoulnar aspect of the metacarpal, again away from the growth plate. The configuration of the tendon graft will then best replicate the UCL.

In older children and adolescents with greater bone stock, the tendon graft can be passed intraosseously. Following exposure of the MP joint, the periosteum of the metacarpal is incised longitudinally proximal to the growth plate on the dorsoulnar surface. A transverse hole is then made large enough to accommodate the tendon graft. Minidrivers with pins are more precise than drills or gouges (see Fig. 43-9). In a small phalanx the tendon graft may need to be split to fit into a small hole in a slender bone. At the distal site in the metaphysis of the proximal phalanx, the periosteum is incised in a dorsopalmar direction. A hole is then made in the same direction, passing from the dorsal aspect of the phalanx to the palmar. The position of the two holes in the proximal phalanx should be such that they are replicating the insertions of the collateral ligament of the joint. The joint is then held with a 0.028-inch C-wire in full adduction and slight flexion. The tendon graft is then passed through the distal tunnel. Proximally the tendon graft is “ink-welled” into the gouge hole in the metacarpal with a transosseous pullout suture or small bone anchor. If a transosseous suture is used this is then tied over a button on the radial side of the thumb.

Arthrodesis or Chondrodesis

The cartilaginous surfaces of both the metacarpal head and the proximal phalanx are exposed through an incision placed dorsal to the midlateral line on the ulnar side of the joint. The cartilage is trimmed down to bleeding bone including the flattened radial side of the metacarpal. Small C-wires are used to position the joint in 20 degrees of flexion with adequate pronation to ensure pulp-to-pulp contact with the index finger. In adolescents and adults all cartilage is removed and a solid bone-to-bone fusion performed. In the adult age group, internal fixation can be used to produce a rigid osteosynthesis.

Opposition (Palmar Abduction) Transfers

The need for opposition transfers can be determined both by assessment of the position of the thumb and observation of the child at play. In a child with an inadequate abductor pollicis brevis muscle, the thumb is adducted with a narrowed first web space. When the child performs pinch tasks, he or she tends to use key pinching maneuvers, because the thumb cannot be abducted adequately to obtain pulp pinch. These children also may use two-handed grasp for larger objects, because they have difficulty with palmar abduction of their thumbs. The degree of first web contracture is a good clinical indicator of the degree of hypoplasia of thumb abductors.

Options

The transfers available for restoration of opposition include the Huber transfer^{47,68,69,71,72} (abductor digiti quinti minimi) or transfer of an extrinsic tendon; the most common being flexor digitorum superficialis to the ring finger.⁷³ (see Fig. 43-9; also see Fig. 43-17). The Huber transfer has the advantages of using a muscle of similar excursion and function to replace the abduction deficiency. It also has the advantage of recontouring the thenar eminence, which is deficient or flattened in these children. The use of this muscle for restoration of thumb abduction may mildly impair the cupping of the palm on the ulnar side of the hand. Some authors prefer the transfer of the flexor digitorum superficialis from the ring or long fingers.^{33,63,74} This transfer has the advantage of also providing tendon slips for UCL reconstruction and the limitation of not improving the contouring of the thenar region (see Fig. 43-10). Rarely, harvesting of an FDS tendon can

result in a swan neck deformity of the donor finger with a lax PIP volar plate or a flexion contracture of the same joint when the tendon has been harvested within zone II of the flexor sheath.

Preferred Method

The Huber transfer can be performed either separately or at the same time as the first web release and UCL reconstruction (see Fig. 43-9). If the transfer is completed in one stage, the first web should be released and the UCL reconstructed with the MCPJ stabilized with a pin, before transfer of the abductor digiti minimi. Our strong preference has been to release the first web space early in life and to delay the Huber transfer and stabilization procedure. At the time of muscle transfer to the radial side of the thumb, it is important to stabilize the ulnar side to counteract this potential deforming force.

Incisions for the transfer include a midlateral incision over the hypothenar eminence at the junction of the glabrous and nonglabrous skin. This incision passes from the metacarpophalangeal flexion crease of the small finger to the distal wrist flexion crease. If necessary, the incision is then extended radially in the normal skin crease at the level of the pisiform bone. In those with very tight skin over the deficient thenar region, a hypothenar skin ellipse can be included as a myocutaneous flap. A second high midaxial incision is centered over the radial side of the thumb MP joint. The hypothenar incision is made and flaps dissected off the muscular fascia. The abductor digiti quinti minimi muscle and its two insertions are identified. The insertion into the extensor mechanism on the ulnar side of the fifth finger is detached before the skeletal insertion into the base of the proximal phalanx. The muscle is then raised retrograde and separated from the more radial flexor digiti quinti muscle. Occasionally, no clear separation exists and both muscles must be raised together. This dissection progresses retrograde toward the pisiform. The neurovascular pedicle enters the muscle on its dorsoradial surface just distal to the pisiform bone and can be clearly identified with careful elevation. The origin of the abductor digiti quinti minimi may need to be released from the ulnar aspect of the pisiform if more length is required. This can be further elevated with a strip of flexor carpi ulnaris if required⁶⁹ (see Fig. 43-9, *C* and *D*). A very generous subcutaneous tunnel is then created for passage of the muscle (or muscles) from the pisiform to the incision on the radial side of the thumb. The muscle is then passed through this tunnel, and both tendons are then reinserted. The shorter tendon from the proximal phalanx is sutured to either the base of the thumb proximal phalanx or radial joint structures, and the longer tendon is interwoven into the extensor mechanism of the thumb. The tunnel must be adequate to allow for postoperative swelling, and the thumb ray must be positioned in full palmar abduction to relieve excessive tension on the transferred muscle tendon unit.⁷¹ The surgeon also has the option of raising this muscle as a myocutaneous flap and placing the skin portion in the lower half of the thenar eminence. In patients with a lax UCL, we prefer to attach the transfer to either the metacarpal head or the radial collateral ligament structures instead of the proximal phalanx.

Interphalangeal Joint Flexion

Although most patients with type II hypoplasia of the thumb have impairment of IP joint flexion, only a few require flexor reconstruction, because the total active range of motion in these patients following web release, MP joint stabilization, and opponensplasty is usually functionally adequate. The ulnar nerve-innervated medial head of the flexor pollicis brevis usually provides strong MP flexion and IP extension. Repositioning the extrinsic extensor of the thumb is required in patients with severe radial deviation of the MP joint, combined with a radially displaced long flexor. The connections between the extrinsic flexor and extensor tendons forming the pollex abductus anomaly are common in this situation and need to be released.⁵⁰⁻⁵² Both of these anomalous tendons are encountered more commonly in type III hypoplasias.

TYPES IIIA AND IIIB THUMB HYPOPLASIA: SEVERE HYPOPLASIA

Anatomy

With type III hypoplasia there is a greater reduction in the size of the thumb, and commonly the origin of the base of the thumb is displaced distally. The distinction between types IIIA and IIIB is made on the presence or absence of the proximal portion of the first metacarpal and a CMC joint⁷⁵ (see Figs. 43-11, 43-13, 43-17, and 43-18). This difference represents a more severe deformity along the continuum of thumb hypoplasia. The length of the osteoarticular column is shorter in type III. The metacarpal is more severely affected than the phalanges and there is an absence of the proximal portion of the metacarpal in type IIIB thumbs.

The radial carpal bones are commonly hypoplastic. The trapezium in type IIIB is usually absent. The distal radius is usually normal in size but the radial styloid process may be absent. The MP joint is completely unstable. The CMC joint is present but hypoplastic in type IIIA and absent in type IIIB. Commonly, there is aplasia of the median nerve-innervated intrinsic muscles and moderate-to-severe hypoplasia of the ulnar-innervated intrinsic muscles: the adductor pollicis and deep head of the flexor pollicis brevis (Fig. 43-11). The radial origin of the first dorsal interosseous from the thumb metacarpal is more severely affected than that from the index metacarpal. The radial head is usually hypoplastic. In many children with type IIIB thumbs it is absent. Extrinsic myotendinous abnormalities are more common than normal anatomy. The FPL can be displaced radially so it acts as an abductor of the MP joint and produces minimal flexion of the first ray, resulting in the so-called pollex abductus.^{47,52} The associated flexor retinaculum of the thumb is poorly developed with either attenuation or absence of the major pulleys of the thumb.

Interconnections between the FPL and the extrinsic extensor are common and contribute to both radial deviation and limitation of IP joint flexion. These radially displaced tendons often attach to anomalous proximal muscle bellies within the palm and do not extend to the normal origin in the forearm. More commonly, the FPL is absent.⁵⁰ Aplasia of the FPL can usually be determined by the presence of some passive IP joint flexion without any active flexion, coupled with an absence of a palmar flexion skin crease at the IP joint level. The long extensor of the thumb and the outcropping muscles (AbPL and EPB) may be absent in type IIIB. The first web is severely constricted.

Management

The management of patients with type III hypoplasia remains controversial. The quandary is one of reconstruction of the deficient IIIB thumb versus ablation and pollicization of the index finger. Cultural, racial, and parental expectations together with the surgeon's experience all play a part in the decision-making process. Reconstruction of all type III thumbs will require:

- First web release and coverage
- Stabilization of the MP joint
- Restoration of opposition
- Extrinsic tendon transfers for extrinsic deficiencies

As a general rule, children with type IIIA thumbs do better with a reconstruction and those with type IIIB thumbs are best treated by pollicization, but factors such as race and cultural beliefs may determine the course of treatment in an individual patient.

Although short, these thumbs are usually of sufficient length to be functionally adequate. Since the advent of free joint transfers, surgeons who are skilled in microsurgical techniques can successfully reconstruct the basal joint of the thumb (see Fig. 43-22). Unfortunately, this procedure does not simulate a normal CMC joint, because a transferred metatarsophalangeal joint of the second toe can only provide motion in a single plane. In addition, extensive tendon transfers

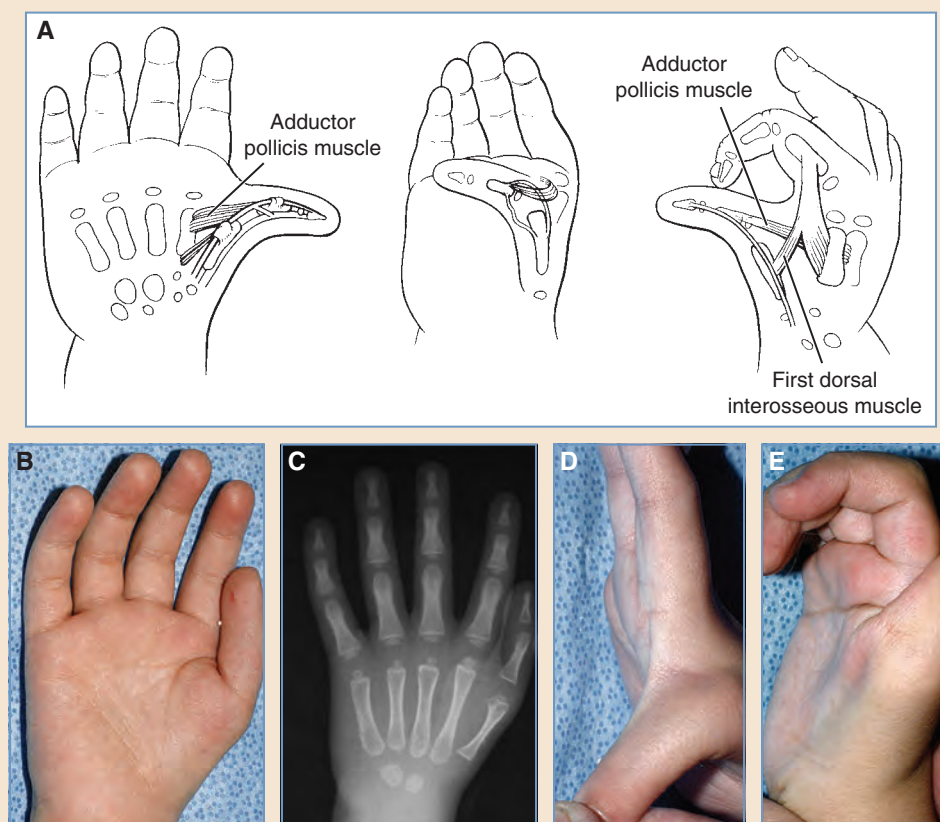


Fig. 43-11 Type IIIA thumb hypoplasia—severe. **A**, These thumbs exhibit more severe skeletal hypoplasia, including the carpal bones. Median nerve-innervated thumb intrinsic muscles are severely hypoplastic or absent, and the ulnar-innervated muscles are present but weak. Extrinsic tendons are abnormal, the “pollex abductus” deformity is frequent, the first web space is small, and the MP joint is unstable (more on the ulnar than the radial sides). The CMC joint and thumb metacarpal are intact in IIIA thumbs. **B-E**, Clinical photographs and a radiograph showing a short, slender metacarpal, an intact CMC joint, severe hypoplasia of the thenar eminence, and a lax MP joint. The absent IP flexion crease is indicative of a deficient or absent FPL

are still required for motion at the MP and IP joints. Those children who have multistaged reconstructions of type IIIB thumbs will also require joint stabilization at one or more levels. More recently, a hemimetatarsal transfer has been described for reconstruction of type IIIB thumbs.⁴⁹ The usual treatment plan for these patients with the more severe type IIIA deformities is to perform the web release and MP joint stabilization as the first stage. Restoration of opposition is then completed as a second procedure with additional extrinsic tendon transfers being done at the same time or as required in additional stages.

Narrowed First Web and MP Joint Instability Options

Release of the contracted first web in patients with type III thumbs is the same as that described for type II hypoplasias. In types IIIA and IIIB the severity of contracture of the first web will determine the type of skin coverage procedure required. Although many children with type III

thumbs are successfully treated with either a four-flap Z-plasty (Fig. 43-12) or a manta ray flap (see Fig. 43-7), those with more severe constriction of the first web need more tissue. Local flap options include: the sliding flap from the ulnar side of the index,⁷⁶ dorsal rotation/transposition flaps with or without skin grafts for the secondary defects,⁶⁴ tissue expansion^{37,65} and reverse-flow forearm flaps⁶⁷ or free tissue transfer.⁷⁷

Stabilization of the MP joint may only require UCL reconstruction. In most patients there is laxity of both collateral ligaments, with the UCL being more affected than the radial collateral ligament (RCL). Construction of the UCL takes precedence but occasionally both ligaments may



Fig. 43-12 Type IIIA reconstruction. **A**, The preoperative appearance of both hands in a youngster born with short stature and floppy thumbs. The thumbs are flail at the MP joint, adducted, and there are no median-innervated intrinsic muscles. **B**, The gross instability at the MP joint precluded any functional pinch. **C** and **D**, At surgery a pollex abductus anomaly was found. After release both collateral ligaments were reconstructed with autogenous tendon grafts anchored into bone. The ulnar collateral ligament of the MP joint is seen on the right. **E** and **F**, The first web space has been released with a four-flap Z-plasty at the same time as an ADQM transfer (Huber). **G**, Buddy taping of the index and long digits was necessary postoperatively to get this child to actively incorporate the use of his new thumbs. The facies of this child is typical of those with associated Fanconi anemia.

require construction. This can be accomplished with the techniques described above in type II hypoplasias. If both collateral ligaments require reconstruction, the same choices are available:

- Reefing of the lax ligaments and joint capsule
- Proximal advancement of the RCL and UCL on the metacarpal
- Tendon graft either as a free procedure or being
- Incorporated as part of a tendon transfer; for example, flexor digitorum superficialis (FDS) from the ring finger, to restore opposition
- MP joint chondrodesis⁷⁰ or arthrodesis; stabilization by chondrodesis or arthrodesis may affect growth in the young child

Preferred Method

In the more severe cases of first web contracture, where coverage cannot be achieved with local procedures, soft tissue coverage is provided by the importation of distant tissue. This can be done with the use of reverse flow island flaps from the forearm or using microvascular free tissue transfer. Both the radial forearm and dorsal interosseous island flaps have been used in congenital hand surgery.^{67,78,79} If a forearm flap is used, the radial forearm flap is preferred, because it is a more robust flap^{80,81} (Fig. 43-13). Because children with preaxial deficiencies may have anomalies

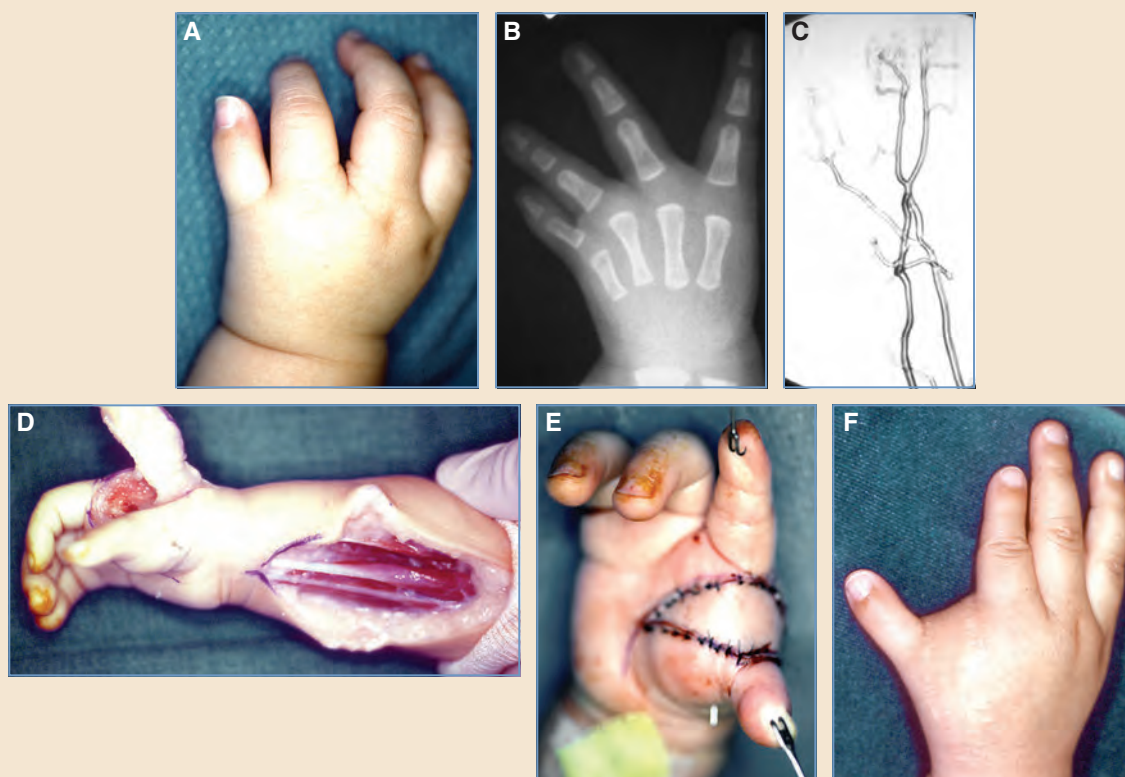


Fig. 43-13 Type IIIA reconstruction with a radial forearm flap. **A**, Preoperative clinical appearance and radiograph showing a small thumb ray with an intact CMC joint and no first web space. **B** and **C**, The angiogram shows a three digits and one thumb hand with communication between the radial and ulnar portions of the hand. **D**, A distally based radial forearm vascular island flap has been passed through a generous subcutaneous tunnel into the defect created by release of the soft tissue between the thumb and index digits. This flap in young children contains a large amount of subcutaneous fat, which cannot be debrided. **E**, There is good perfusion after a tension-free inset into the new web space. **F**, One year later, the first web space has been well maintained by this full-thickness flap.

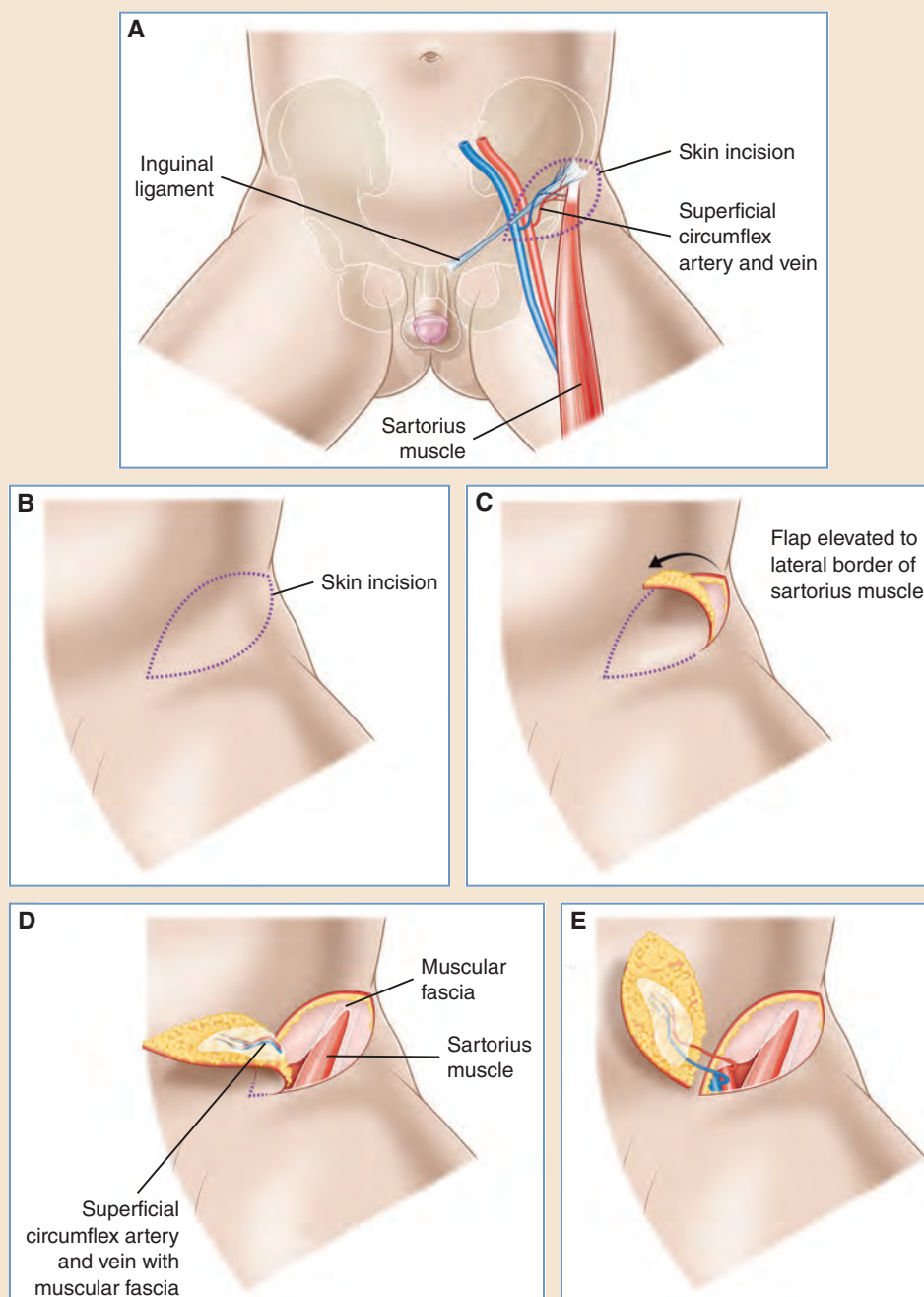


Fig. 43-14 Technique for elevation of a groin flap. **A**, Skin markings for the flap are drawn based on the axis of the superficial circumflex iliac artery (SCIA), which runs parallel and inferior to the inguinal ligament. **B**, A skin island is designed to permit primary closure of the defect. **C**, Elevation begins laterally in a suprafascial plane. **D**, At the lateral border of the sartorius muscle, the plane of dissection deepens to include elevation of the deep fascia. The SCIA can be visualized through the fascia. **E**, The vessels are dissected to their origin from the common femoral artery and the femoral vein (occasionally the superficial circumflex iliac vein originates from the deep circumflex iliac vein). Flap perfusion should then be assessed, because a prominent superficial venous system mandates additional anastomosis to augment outflow of the medialmost skin.

of the radial vasculature, preoperative angiography of the forearm and palmar arches is essential. When the palmar arches are incomplete, the radial forearm option cannot be used.

If the treating surgeon is experienced with pediatric microsurgical techniques, the use of free tissue transfer is an excellent choice for resurfacing the first web in these children. Our preferred transfer is the groin flap because of its donor site and relative thinness. The radial artery is the usual arterial recipient and a dorsal vein for the venous anastomosis (Fig. 43-14). Sliding and rotation flaps from the visible dorsal portion of the hand with skin grafts in the donor region are popular with many surgeons, but may produce unacceptable scarring on the socially presented surface of the hand. Dorsal skin expansion is another option for those experienced with these techniques⁶⁴ (Fig. 43-15).

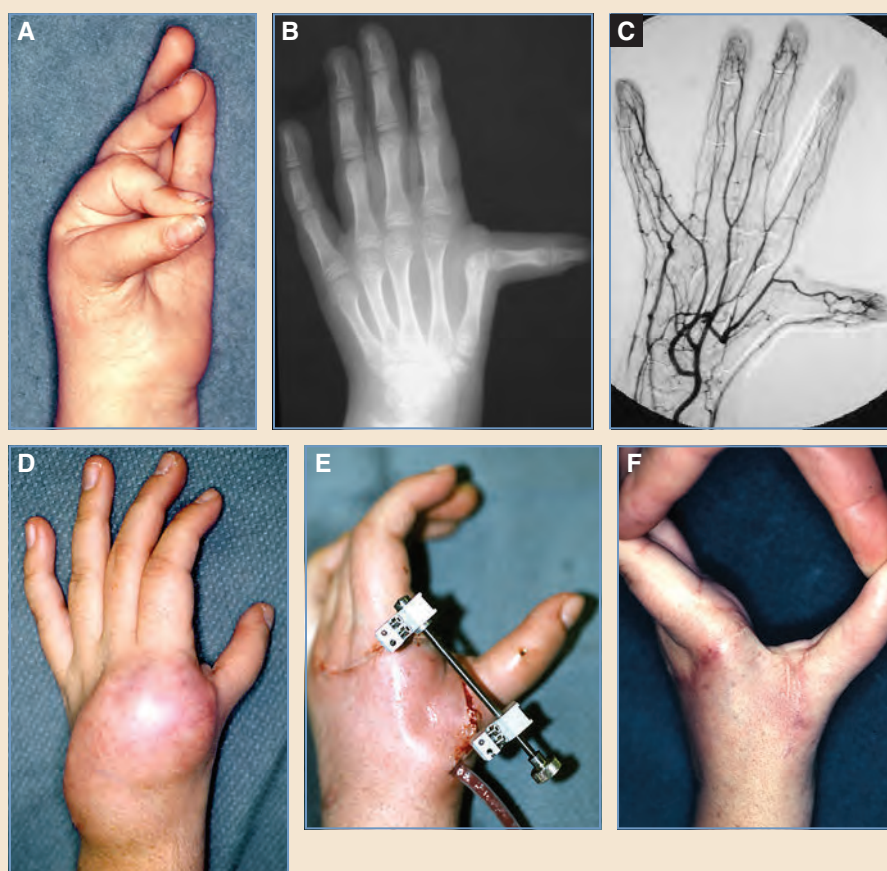


Fig. 43-15 Skin expansion for the first web space. **A**, This 10-year-old child with type IIIA thumb hypoplasia lacks any stability with pinch as a result of an attenuated ulnar collateral ligament. A Huber transfer performed earlier in childhood pulled the thumb into radial deviation. **B**, The radiograph shows a 90-degree deviation at the MP joint. **C**, An inadequate arterial communication between the radial and ulnar sides of the hand ruled out a distally based forearm flap reconstruction of the first web space. **D**, Dorsal skin has been expanded over a 4-week period. **E**, This skin was advanced to provide adequate coverage. At the same time, the ulnar collateral ligament was reconstructed with both local tissue and a free tendon graft. **F**, Following reconstruction, the patient has a stable MP joint and a wide first web space lined by full-thickness tissue.

The thumb-index web space is released with a longitudinal skin incision centered over the middle of the web on both the palmar and dorsal surfaces. This incision will adequately expose the UCL of the MP joint. A separate radial midaxial incision on the other side of the thumb is required to reconstruct the radial collateral ligament (RCL). In young children we often choose to simply imbricate the collateral ligaments of the MP joint. In thumbs in which the UCL is more severely affected than the RCL, a tendon graft of the UCL with imbrication of the RCL and dorsal capsule is a good option. Bilateral tendon grafts are rarely used but may be required in very severe cases. Chondrodesis is another option. Following MP joint reconstruction the MP joint is held in position with a 0.028-inch C-wire for 3 weeks. A second larger threaded C-wire may be needed transversely between the first and second metacarpals to hold the released ray abducted and pronated. Individual pins attached to an external fixation device will accomplish the same (see Fig. 43-15).

Following release of the web, a diamond-shaped defect is created. A template is made of this defect and transferred to the groin, or the volar forearm overlying the course of the radial or the dorsal proximal forearm over the posterior interosseous artery, which has been mapped out by Doppler examination before tourniquet inflation. If a forearm flap is being used, the pivot points for the radial forearm and dorsal interosseous forearm flaps are the radial styloid and the proximal edge of the extensor retinaculum, respectively. The choice of which flap is determined before the surgery with the use of angiography. It is important to elevate both of these flaps with a 1.5 to 2.0 cm cuff of fascia beyond the skin paddle, because in this young age group cutaneous perforating vessels to the skin flap can be quite tenuous. The flaps are raised retrograde by routine methods, as previously described^{67,78-82} (see Fig. 43-13). No attempt should be made to thin these flaps before inset, because this can jeopardize their vascularity. The flaps are tunneled into the defect while directly observing the pedicle, which cannot be kinked or rotated. The flap is inset using 5-0 and 6-0 mild chromic catgut, with the forearm wound being closed with a subcuticular suture. If arterial flow is sluggish after the tourniquet is released, the flap may be “supercharged” by a direct arterial anastomosis to the radial digital artery of the index finger.

When using a free groin flap, the surgeon needs to consider the harvest site to avoid future hair growth on the flap.⁷⁷ The flap is raised in a retrograde fashion (see Fig. 43-14). Meticulous technique is required to avoid damage of the vasculature to the flap. Intimate knowledge of the anatomy and anatomic variations is required. The size of the SCIA in young children is less than 1 mm, and this technique should only be undertaken by experienced pediatric microsurgeons (Fig. 43-16).

Opposition Transfers

In patients with type IIIA and IIIB thumb hypoplasia, the preferred method for restoration of opposition or palmar abduction is the Huber transfer (see type II opposition transfers). Occasionally, in older children and adolescents, the use of an FDS tendon is quite practical, because this method provides simultaneous opposition transfer and collateral ligament reconstruction. In thumbs with severe laxity of both collateral ligaments of the MP joint but only mild first web contracture, a single-stage procedure can be performed: web release with a manta ray flap (see Fig. 43-7), and FDS transfer for opposition (Fig. 43-17). If there is severe instability of both sides of the MP joint associated with a very contracted first web, a two-stage procedure is preferred: web release with distant flap reconstruction with a free groin flap (see Fig. 43-16) or reverse-flow forearm flap repair, followed by FDS transfer with bilateral ligament reconstruction.

Harvest of the FDS tendon to the ring finger is done through a transverse incision in the distal palmar skin crease. The A1 pulley of the ring finger is decompressed and the FDS tendon identified. With the ring finger pulled into flexion, a right-angle retractor is used to pull the FDS

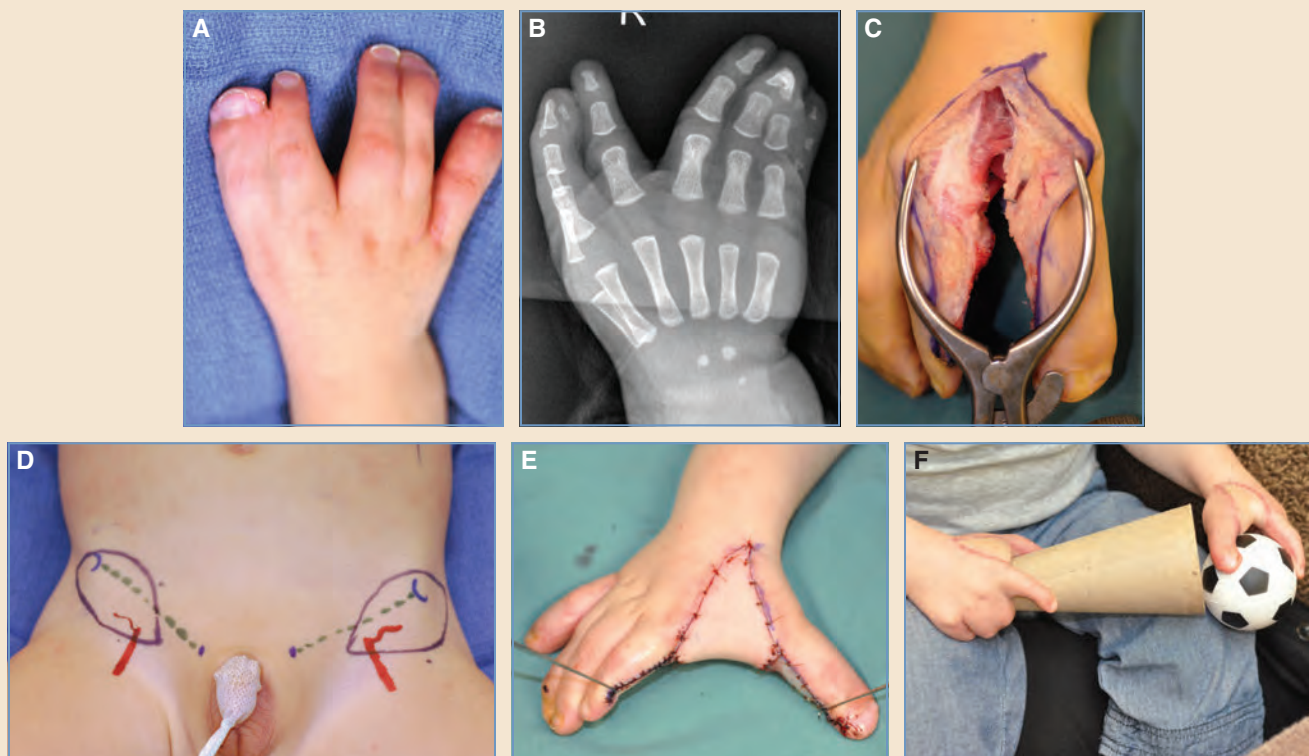


Fig. 43-16 A child with thumb/finger polysyndactyly, with first web syndactyly treated with bilateral free groin flaps. **A**, Preoperative appearance, and **B**, radiologic findings. **C**, Intraoperative release of the first web. **D**, Bilateral groin flaps planned. **E**, Groin flap inset into right first web. **F**, Postoperative appearance of the hands.

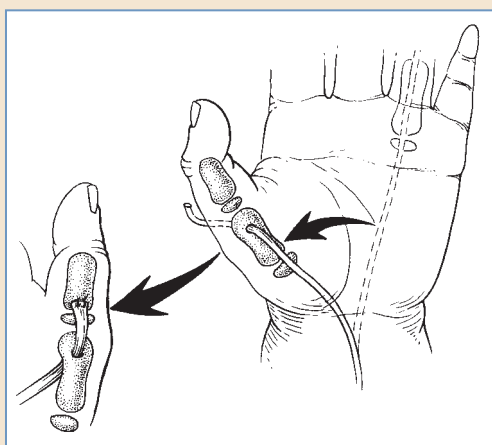


Fig. 43-17 FDS dual transfer. The preferred technique for individuals with flail thumbs lacking good thenar muscles is the transfer of the ring flexor digitorum superficialis to the radial side of the thumb. The tendon is passed from whatever direction the surgeon chooses from the forearm. One slip is passed through a gouge hole proximal to the metacarpal head, and the tendon is secured tightly. The two tendon slips can then be used for construction of collateral ligaments on both sides of the joint.

proximally. Both slips of the FDS are incised as distal as possible. Through a longitudinal incision on the radial side of the flexor carpi ulnaris (FCU), the disconnected FDS tendon is delivered. The tendon should not be allowed to desiccate beneath the hot lights. A pulley is then constructed from part of the FCU and a generous subcutaneous tunnel is made from the wrist incision to a radial midlateral incision centered over the MP joint of the thumb.

We have encountered secondary ulnar nerve compression neuropathies when the transfer has been directed through Guyon's canal. The tendon is then tightened and split longitudinally to a point at which the full tendon meets the radial side of the metacarpal head. In patients with enough diaphyseal bone, the radial slip of the FDS tendon is passed through the metacarpal head to the ulnar side of the joint, which has been exposed through the first web release incision or through a midlateral incision. The UCL is constructed with one slip and the RCL with the other.

Attenuated sagittal fibers of the extensor mechanism and dorsal capsule are imbricated and tightened at the same time. Before both slips of the FDS are completely sutured in place, it is important to assess the motion of the MP joint so that the tendon grafts do not restrict this motion. Since these reconstructed ligaments loosen with time, increased tension and decreased motion are preferred initially.

Interphalangeal Joint Motion

The majority of patients with type III hypoplasias of the thumb lack IP joint flexion as a result of hypoplasia or aplasia of the flexor pollicis longus myotendinous unit. This anomaly gives these thumbs their characteristic slender appearance and associated lack of flexion creases. In this group it is unusual to have absence of both the FPL and the deep head of the FPB, which usually accounts for some thumb flexion. Tendon transfer for IP joint flexion should only be done in patients who have greater than 30 degrees of passive IP joint flexion and a stable joint.

The absence of both short and long flexors (FPB and FPL) will severely impair both pinch and grasping functions. In patients who do not have the pollex abductus deformity, the pulley system is significantly attenuated, but the FPL tendon is usually more developed. Transfer of aberrant tendons into normal positions should not be done, since these tendons are commonly connected to poorly developed proximal muscle bellies. The results of these transfers are predictably poor.

Reconstruction of type IIIA thumbs that lack IP joint flexion is best accomplished in two or more procedures. In a patient with the pollex abductus anomaly, the abnormal FPL is divided and any flexor-extensor interconnections released at the time of first web release and MP joint stabilization. The FPL tendon is then reinserted into the base of the first metacarpal to function as a long abductor of the thumb. Pulley reconstruction will be required before transfer of ring or middle finger FDS and can be done at the next stage, in conjunction with the Huber transfer for opposition. The distal portion of the malpositioned FPL can be used for pulley reconstruction. The placement of a silicone tendon rod at this stage produces a tunnel for the ensuing tendon transfer. Transfer of the FDS tendon for FPL is then done at a third stage. In patients who have aplasia of the FPL but a reasonable pulley mechanism, restoration of active IP joint flexion can be performed by transfer of an FDS tendon at the same time as the Huber transfer.

Thumb extension may also be absent in the more severe thumb hypoplasias. The extensor indicis proprius is usually available for transfer. Commonly there is a satisfactory distal extensor tendon present, which can be used and obviates the need for a tendon graft. If this is also absent then an extensor communis tendon can be used and, following transfer, thumb extension will occur simultaneously with digit extension. Other transfers such as FDS to EPL, brachioradialis or extensor carpi radialis longus plus a tendon graft to EPL can be considered. The reconstruction of those thumbs with absent extrinsic extensors follows the same algorithm as that for the treatment of the clasped thumb.⁸²

TYPE IIIB THUMB HYPOPLASIA

Options

The management of type IIIB hypoplastic thumbs is more controversial. Reconstruction in these patients will require multiple procedures, including stabilization of the base of the thumb ray. These thumbs are diminutive, usually arise quite distally in the hand and have multiple intrinsic and extrinsic deficiencies (Figs. 43-18 and 43-19). Because of the size of the thumb, reconstruction may appear to be an attractive option. Many parents are often reluctant, some strongly insistent, about considering any option other than reconstruction. In any reconstruction of a type IIIB thumb, the surgeon must be able to provide a mobile, stable thumb of sufficient length and to preserve growth potential with functional intrinsic and extrinsic myotendinous units and an

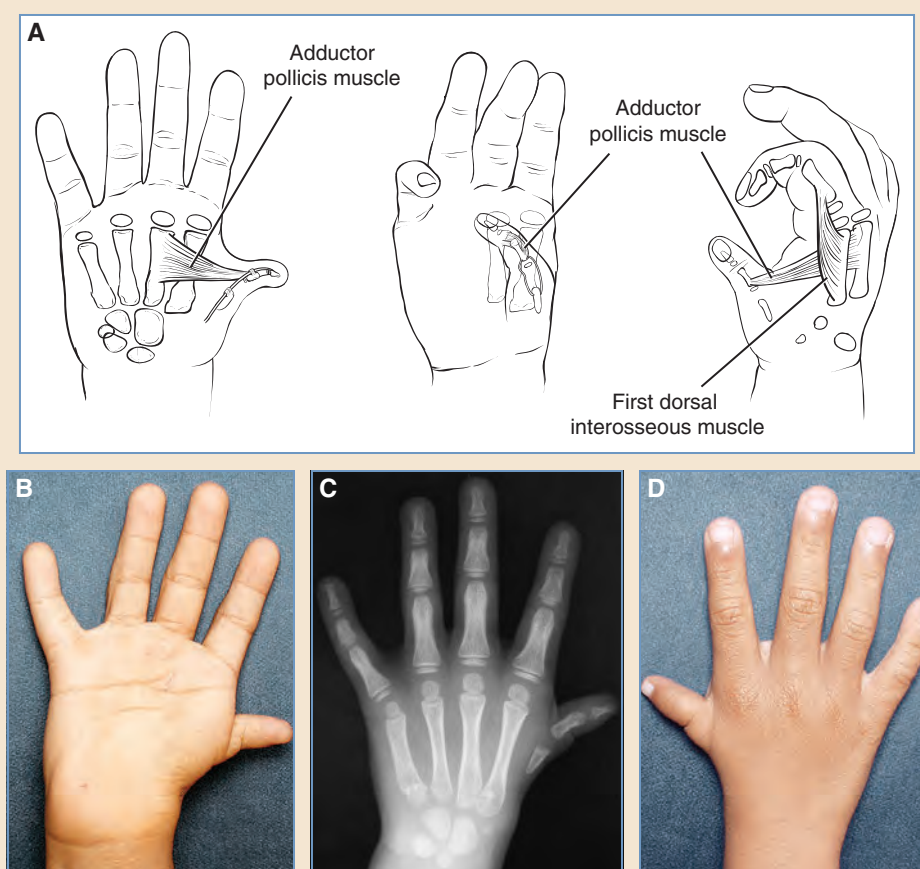


Fig. 43-18 Type IIIB thumb hypoplasia. **A**, The median nerve–innervated thenar intrinsic muscles are absent entirely. There is severe hypoplasia of the AddP, the FPB-lateral head, and the ulnar origin of the first DI. The CMC joint articulation of the proximal metacarpal is absent. The MP joint is very lax or absent on both radial and ulnar sides. Flexor and extensor extrinsic tendons are severely hypoplastic or absent. **B-D**, Clinical appearance and radiograph of a 4-year-old patient with a well-formed but functionless floppy thumb. There is no skeletal stability or extrinsic flexion or extension. Very small flexor or extensor tendons may be present and can result in some movement.



Fig. 43-19 Spectrum of metacarpal and CMC deficiencies. **A-D**, There is no arbitrary distinction between specific categories of thumb hypoplasia based on the skeletal appearance alone. These thumbs, which show varying degrees of hypoplasia with a presumably intact CMC joint, would be classified as type IIIA. **E-H**, The smaller metacarpals in these examples would signify type IIIB thumbs. **I-L**, These thumbs, with no skeletal connection, would be classified as floating thumbs, type IV. When a tight syndactyly exists between the thumb and index digit, an associated congenital heart defect (Holt-Oram syndrome) may be present.



Fig. 43-20 Bypass of use of the thumb in preference for second web space grip.

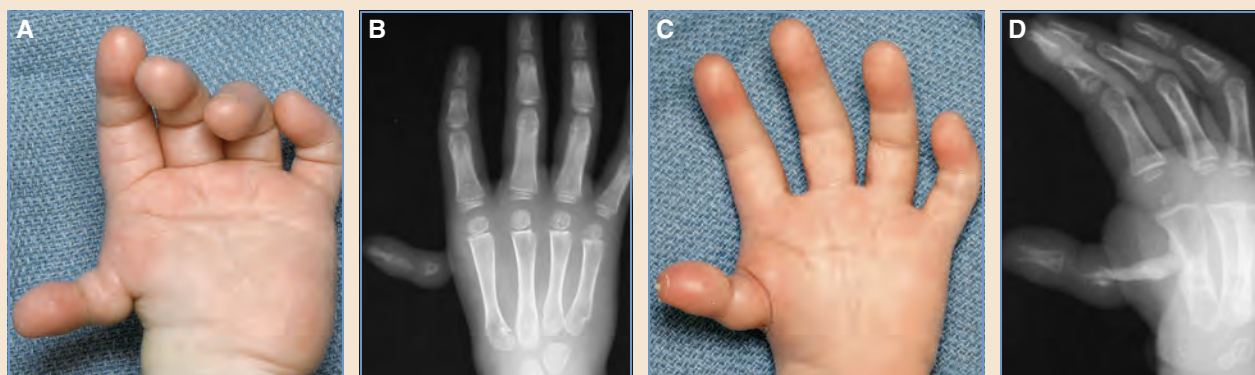
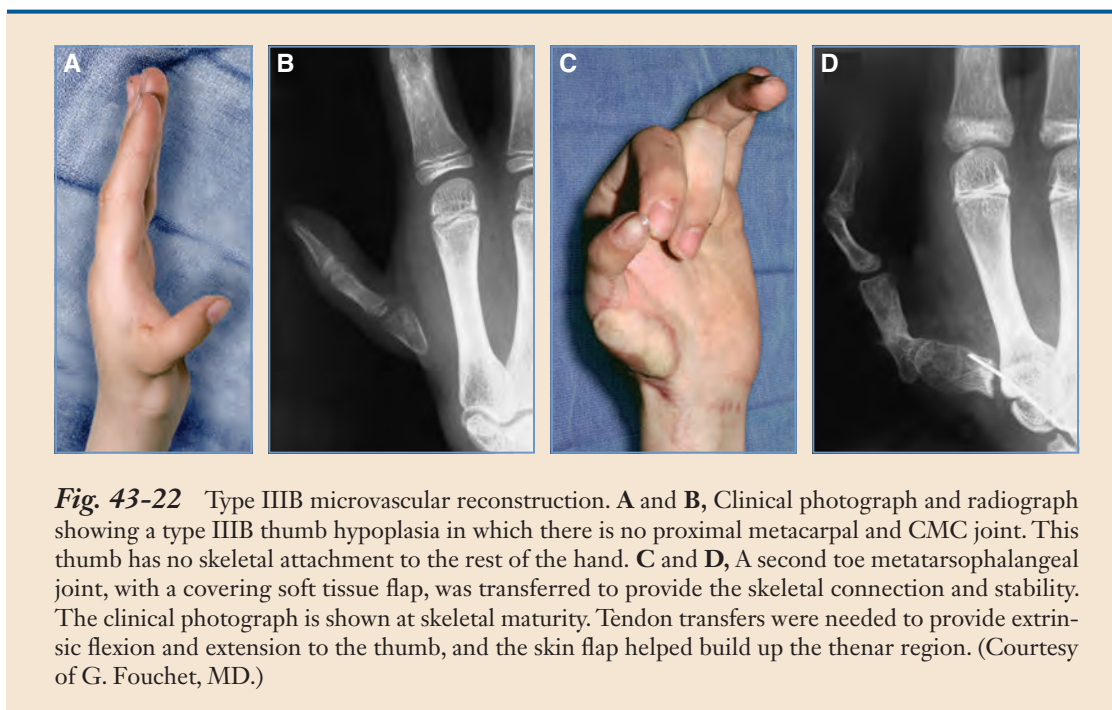


Fig. 43-21 Type IV thumb construction. **A** and **B**, The preoperative radiograph and clinical appearance of a type IV thumb in a child whose parents insisted on saving it. **C** and **D**, A bridging iliac corticocancellous bone graft was used to stabilize the thumb, which became an immobile post and did not grow commensurately with the child. At 14 years of age, the patient came to our clinic and asked when I would give her a “normal” thumb.

adequate scar-free first web space. Unfortunately, it is common for reconstructed thumbs in these patients to be stiff and unaesthetic, and the reconstructed thumb will still be bypassed in preference to the index finger. Observation of the child at play will usually direct the surgeon and family to the best course of management (Fig. 43-20). A child who bypasses the hypoplastic thumb before reconstruction will usually continue to do so after reconstruction.

Staged Reconstruction

The order of procedures in the reconstruction of type IIIB hypoplastic thumbs is (1) stabilization of the thumb osteoarticular column combined with first web release, (2) MP joint stabilization and opponensplasty, and (3) staged extrinsic tendon transfers as required, FDS for FPL and EIP for EPL. Adductorplasty may also be required (Fig. 43-21).



Using microvascular free tissue transfers, surgeons now have the ability to transfer joints with the growth plates of the accompanying bones. The procedure of choice for stabilization of the base of the flail-type IIIB thumb is the transfer of the metatarsophalangeal joint of the second toe (Fig. 43-22). Dorsal skin with veins is included for augmentation of the thenar region. The toe MP joint is positioned in hyperextension at the base of the thumb with a small portion of the distal diaphysis with the metatarsal head. Following this microvascular transfer, the reconstruction is similar to that described previously for a type IIIA hypoplastic thumb. Other options for the type IIIB thumb exist. Hemimetatarsal transfer of the second metatarsal⁴⁸ or a nonvascularized iliac crest graft may be used to provide a nonmobile strut connected to the base of the index metacarpal; however, iliac crest used as a bone graft may resorb, will not grow, and is not recommended.

Rarely, a type III thumb may present with absence of the diaphysial portion of the metacarpal and an intact CMC joint. In this instance an intercalated iliac crest bone graft is the treatment of choice. Despite limitations, staged reconstructions of type IIIB thumbs may result in high parent and patient satisfaction. Long-term follow-up of a limited number of these patients reveals limited function in patients who are nevertheless very enthusiastic about their thumbs.

Preferred Management

Even though cultural and parental beliefs can provide great pressure for reconstruction in this group of patients, we believe that a well-executed pollicization of the index finger will provide a superior result to staged reconstruction. We have had adequate experience with both methods. This recommendation is of greater significance for the bilateral deformity.

TYPE IV THUMB HYPOPLASIA: POUCE FLOTTANT

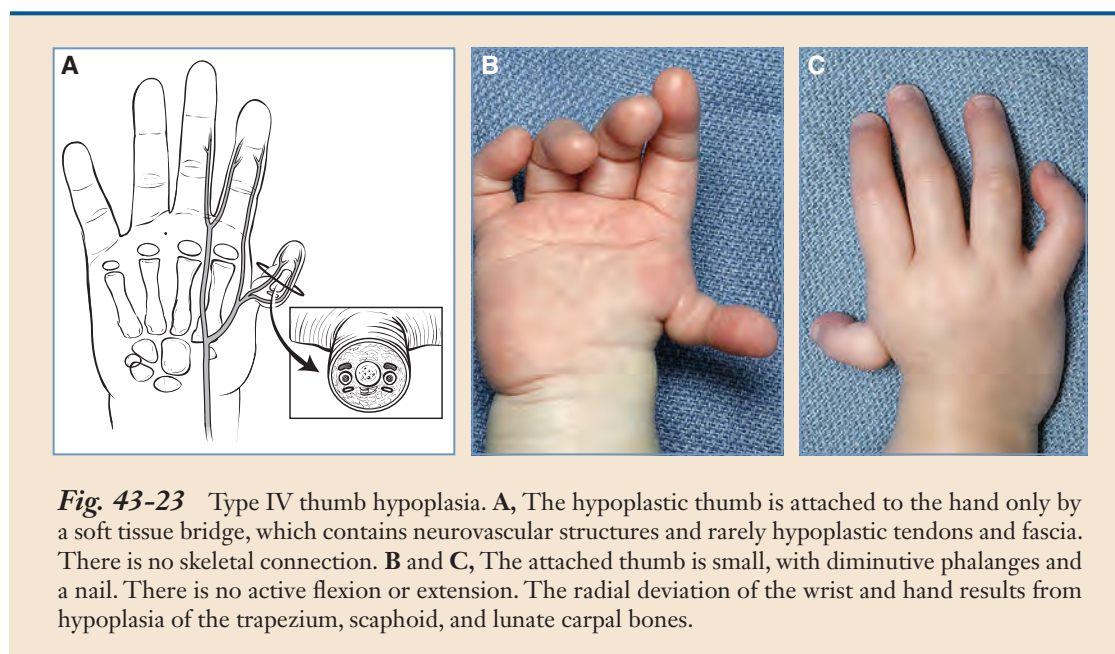
Anatomy

Pouce flottant is the French term for “floating thumb.” Such thumbs arise distally from the palm and usually lie along its radial midaxial border. These diminutive thumbs are attached to the hand only by a soft tissue pedicle, which has been described by Littler⁸³ as “nature’s own neurovascular pedicle” because of the presence of a single digital nerve and artery within the skin bridge (Fig. 43-23). These thumbs do not have a metacarpal bone. One or two small phalanges tend to be present within the soft tissue envelope of the thumb. If two phalanges are present, the IP joint does not have passive motion. The intrinsic muscles to the thumb are absent, as is the radial head of the first dorsal interosseous muscle. The ulnar origin of the first dorsal interosseous muscle is usually present, and this muscle functions as an abductor indicis.

Occasionally, motion is seen in a *pouce flottant* because of the presence of very rudimentary tendon units passing into the hypoplastic digit. Exploration of these thumbs reveals fascial slips passing through the pedicle that merge with the fascial bands within the floating digit. Within the carpus the trapezium is absent, and less commonly, the scaphoid. The trapezoid may be hypoplastic. The radial styloid may be absent, but the distal end of the radius is normal in most of these children.

Management

The best reconstruction for type IV thumbs is ablation and pollicization of the index finger. The larger variants of type IV thumbs with skin bridges of 1.0 cm or larger often have well-developed thenar intrinsic muscles that are quite effectively transferred to improve both palmar abduction and adduction of the pollicized index ray.



Staged construction of a thumb using the pouce flottant has been described using the methods outlined for reconstruction of a type IIIB and IV hypoplastic thumbs.^{49,84} The final results of these staged procedures are comparable and often the salvaged pouce flottant is stiff, immobile and “gets in the way” in an abducted position from the rest of the hand (see Fig. 43-21). Stability can be obtained but motion is severely impaired. Because these thumbs are small and have a rudimentary nail complex, the constructed thumb will definitely be less aesthetically acceptable than a pollicized index finger.

TYPE V THUMB HYPOPLASIA: APLASIA

Anatomy

In type V there is absence of the thumb osteoarticular column and the associated intrinsic and extrinsic myotendinous units. There is aplasia of the radial origin of the first dorsal interosseous muscle and in 50% of cases the entire muscle is absent (Fig. 43-24). The trapezium and commonly the scaphoid are aplastic, with the trapezoid being hypoplastic, and the collective carpal hypoplasia results in a radial deviation of the hand and wrist. The radius may be normal, hypoplastic, or absent. When the radius is present, the radial head is often subluxed or dislocated at the elbow level. With aplasia of the thumb the index finger tends to “autopollicize” (see Fig. 43-24). The pulp of the index finger widens and the digit pronates and sits in an abducted position resulting in a widening of the intermetacarpal space and attenuation of the intermetacarpal ligament. At best this is a poor substitute for normal key pinch and thumb index grasping maneuvers.

In more severe degrees of radial dysplasia (preaxial failure of formation) the index finger may also be hypoplastic, joined to the long finger by an incomplete syndactyly, and demonstrates markedly reduced active and/or passive range of motion at the MP, PIP, and DIP joint levels. With associated hypoplasia of the radius, the index and long fingers are rarely normal.

Management

With severe hypoplasia (types IIIB and IV) and aplasia (type V) of the thumb, the preferred treatment is pollicization of the index finger. Although reconstruction of these diminutive thumbs is feasible and preferred in some societies because of the importance placed on a full complement of fingers, we think a well-executed pollicization will predictably result in a more functional and aesthetic thumb.

Pollicization

Gosset⁸⁵ and Hilgenfeld,⁸⁶ working independently, designed methods of transposition of the index finger into the thumb position. All of their cases involved posttraumatic deformities in which the proximal thumb metacarpal and CMC joint were intact. Littler⁸⁷ studied their work, refined their techniques, developed new incisions, and expanded their basic concept to include the congenitally absent thumb. His unique and detailed drawings popularized the concept of index transposition among hand surgeons. Buck-Gramcko^{88,89} in Germany contributed further refinements during an extensive experience with radial dysplasias associated with the thalidomide crisis in western Europe and elevated this operation to its present status.

Although pollicization greatly enhances the function of these hands, it is important to remember that the index finger is only placed in a more strategic position to simulate a thumb. The newly created CMC joint was the index MCP joint and therefore does not have the same degree of freedom as the normal CMC joint of the thumb.

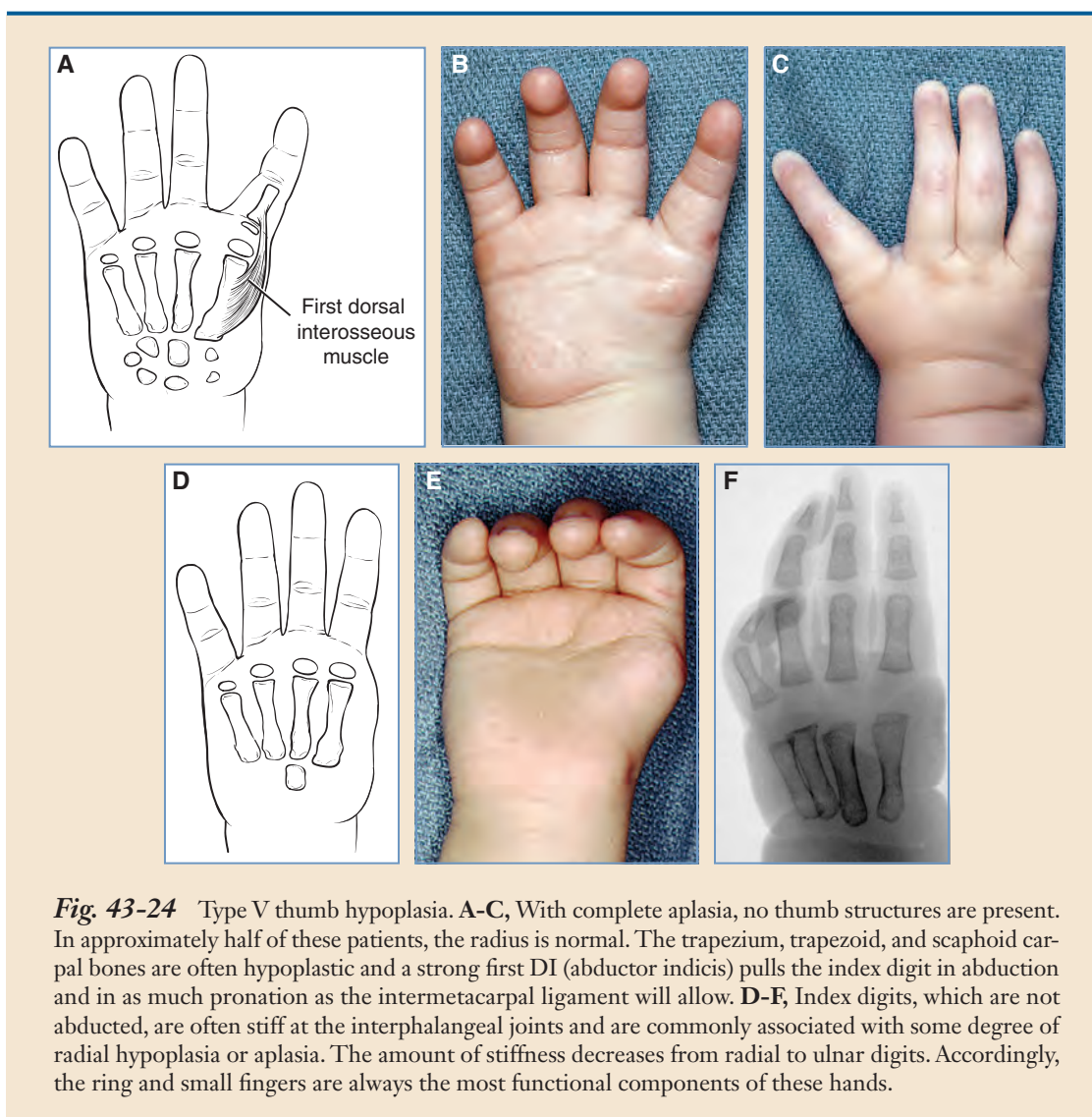


Fig. 43-24 Type V thumb hypoplasia. **A-C**, With complete aplasia, no thumb structures are present. In approximately half of these patients, the radius is normal. The trapezium, trapezoid, and scaphoid carpal bones are often hypoplastic and a strong first DI (abductor indicis) pulls the index digit in abduction and in as much pronation as the intermetacarpal ligament will allow. **D-F**, Index digits, which are not abducted, are often stiff at the interphalangeal joints and are commonly associated with some degree of radial hypoplasia or aplasia. The amount of stiffness decreases from radial to ulnar digits. Accordingly, the ring and small fingers are always the most functional components of these hands.

Timing

There is no consensus on the best age at which to perform a pollicization. We have performed this procedure as early as 4 months of age and as late as 15 years of age. Those in favor of early surgery feel that because the child develops thumb awareness by 3 months of age and prehension by 1 year, it is important to place the index finger in the thumb position by 1 year so the child can develop a “thumb.” There is no cortical representation of the absent thumb, and the index finger is the most radially represented digit in the cortex. Therefore early surgery will take advantage of “cerebral plasticity” and help obviate the need for the child to “relearn” using the newly positioned index finger. Edgerton et al⁴⁵ suggested that early surgery is also advantageous to the developing child’s body psyche, because a child is aware of his or her hands early, and corrective surgery may lessen the perception of an unfavorable body image. Proponents of late

surgery feel that with a larger hand and less subcutaneous fat, a much more precise procedure can be accomplished. The timing of pollicization needs to be determined by the treating physician, because performing this technically demanding surgery on small hands requires meticulous attention to detail and technique. The tyro in hand surgery with limited experience is cautioned to avoid early pollicizations.

Preferred Management

Incisions

Before inflation of a pneumatic tourniquet, the incisions are marked.^{87,90} A racquet-shaped incision is made at the base of the index finger just proximal to the digitopalmar flexion crease, with a radial extension that continues into the palm toward the new thenar flexion crease (Fig. 43-25, *A* through *C*). The palmar incision ends at the base of the index metacarpal. The rotation-recession movement of the index digit then invites a Y-V advancement of tissue from the radial border of the hand into the radial, mid-dorsal or ulnar surface of the transferred index ray. The exact placement of this incision depends on the tissue available and is not made before the index has been moved. Once the index digit has been moved and the metacarpal head fixed into position, a backcut incision is made on the dorsal surface of the index proximal phalanx (now the thumb metacarpal). Dorsal and ulnar skin flap from this digit is then rotated into the new first web space to create a more aesthetic web, which extends to the MP joint of the new thumb (formerly the PIP joint of the index finger).

The basic plan of the incisions for pollicization in the presence of a type IIIB or type IV (pouce flottant) hypoplastic thumb is not greatly altered. It is best to plan incisions first as though no thumb remnant is present and then to see how one can incorporate extra tissue into the original design. The level of hypoplastic thumb placement along the index ray, as well as the amount of tissue available, varies tremendously. Often in the Holt-Oram syndrome (congenital heart disease and radial dysplasia) a hypoplastic thumb is joined to a normal index ray by a complete simple syndactyly. In these cases, the thumb is excised through a midaxial radial incision at the same time as pollicization, but this can be performed as a separate procedure.

Soft Tissue Dissection

After meticulous marking, limb elevation, and exsanguination, the incision is made around the base of the index finger and into the palm. The dorsal skin flap is raised in an areolar plane superficial to the dorsal veins, lymphatics, and sensory nerves (Fig. 43-25, *D*). Palmar skin is elevated superficial to the palmar fascia. If present, the skeleton of the hypoplastic thumb is filleted from its soft tissue envelope and present intrinsic muscles are individually isolated and dissected to their proximal origin. The palmar fascia is then incised parallel to the index extrinsic flexor tendons, and the A1 pulley is incised in its volar midline (Fig. 43-25, *E*). The superficial palmar arch is identified and the vessels to both sides of the index ray are identified. The presence of fat cells and pacinian corpuscles often make the identification of neurovascular structures easier. The common digital artery to the index-long web space is followed to its bifurcation, and the radial digital artery to the long finger is ligated (Fig. 41-25, *F*). The radial digital nerve to the long finger is identified and separated proximally from the common digital nerve by fascicular dissection (Fig. 43-25, *G*). Neural and vascular loops are common on either side of the index ray and when noted should be dissected under appropriate magnification. The transverse metacarpal ligament is next incised (Fig. 43-25, *H*). Dorsal veins and nerves are not individually isolated from their dorsal soft tissue pedicle. Commonly, the major venous drainage from the index finger may join a large vein over the long metacarpal before it courses proximally. Dissection must then extend over the adjacent long metacarpal (Fig. 43-25, *I*).

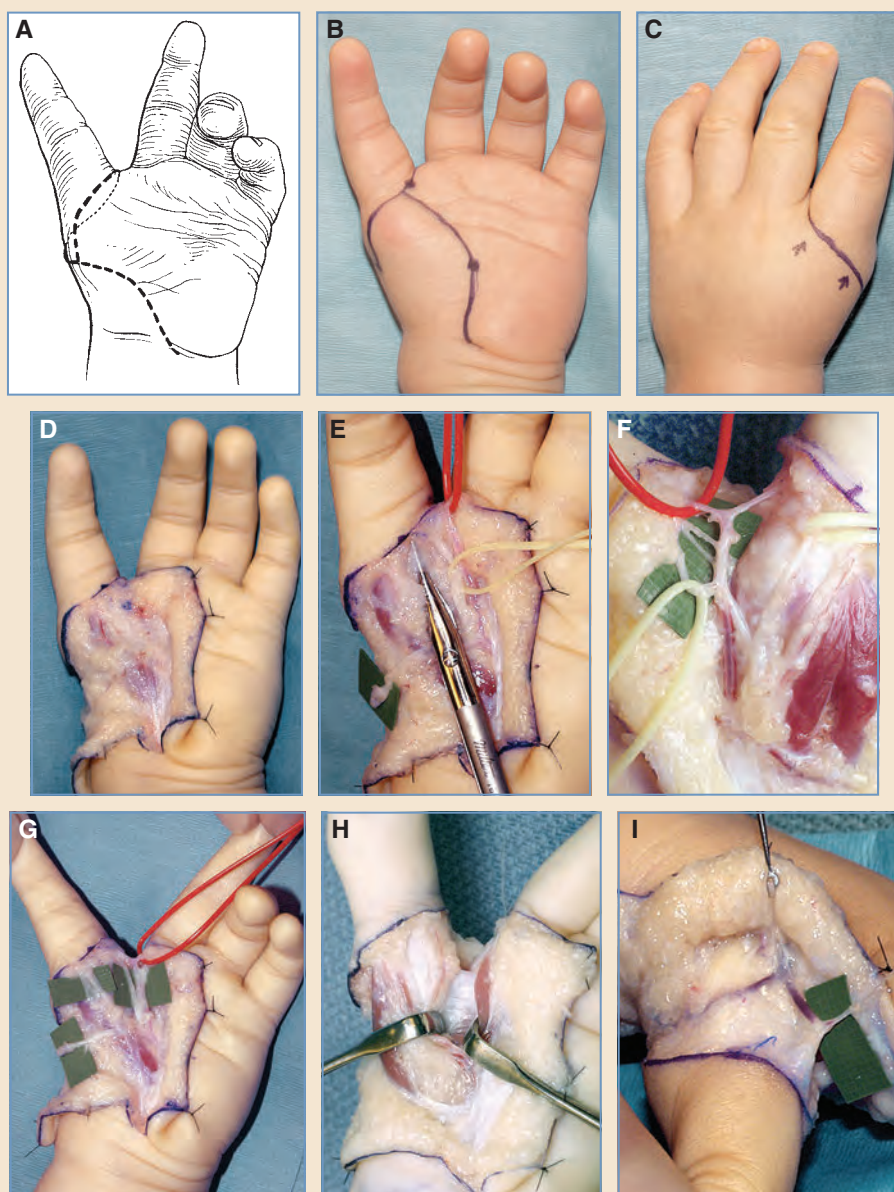


Fig. 43-25 Pollicization technique. **A-C**, The illustration and palmar view show the plan for the incisions. The most important marking is the location of the future thenar flexion crease within the palm. The palmar base of the index finger will be the midportion of this flexion crease. The dorsal marking is made and the visible dorsal draining veins are marked by arrows. **D**, The palmar flaps are elevated above the palmar aponeurosis. **E**, A full release of the first annular pulley (palmar to the scissor tips) is completed. **F**, A neural loop around the common vessels to the web space must be carefully teased apart back to the level of the palmar arch. **G**, The common digital bundle to the index–long finger web space is isolated. The arborization to the radial side of the long finger (*red loop*) will be ligated. Note the neuroma in the dissected sensory nerve to the floating thumb, which was ligated in the newborn nursery. **H**, The transverse metacarpal ligament is exposed before its transection, which then provides increased mobility of the ray for the intrinsic muscle dissection. **I**, The dorsal venous system is easily exposed by scissor dissection between the two layers of dorsal fat/areolar tissue.

The interosseous muscles to the index finger are next identified. The palmar interosseous is consistently present on the ulnar side, whereas its dorsal interosseous partner on the radial side is absent in 50% of patients with thumb aplasia. Occasionally, the first dorsal interosseous muscle is found to have two muscle bellies. The distal skeletal and tendinous insertions need to be dissected carefully so that 1.0 to 2.0 cm of tendon is available for reattachment. These intrinsic muscles are then raised from distal to proximal toward the middle third of each muscle. Their metacarpal periosteal muscle origin remains intact and is saved and distal free periosteum is discarded, because it often becomes the site of ectopic bone formation.⁹⁰ The lumbrical of the index finger is not dissected (Fig. 43-25, *J* through *L*).

The extensor indicis proprius and extensor digitorum communis tendons to the index finger are then divided proximal to the extensor hood and reflected. These extensors require significant shortening before reattachment to the newly positioned index finger. Some shorten the extrinsic flexor tendons.⁹¹ This is best done in patients with stiff index fingers and those with associated radial dysplasia, because this will result in improved grip strength in those patients.⁹² This is done following osteotomy and repositioning

Osteotomy and Repositioning

Once the soft tissue dissection has been completed, the index metacarpal is exposed dorsally, and subperiosteal dissection is completed from the metacarpal base to the epiphysis (Fig. 43-25, *K* and *L*). Intrinsic muscle attachments to the periosteum are left intact. A transverse osteotomy is made through the epiphysis, and the collateral ligaments and palmar plate to the MP joint are left intact. With deep retraction, the metacarpal base is cut obliquely to serve as a pillar to support the index metacarpal head, which will be positioned anterior to it (Fig. 43-25, *M*). The metacarpal head, as part of an ovoid joint, becomes the trapezium as a substitute for the toroidal or universal saddle joint.

The shaft of the second metacarpal is removed and the index digit is both recessed and rotated into the thumb position. The metacarpal head is hyperextended and fixed volar to the base of the index metacarpal so that an appropriate metacarpal arch is created to simulate that of the normal thumb.^{90,93} This maneuver will orient the new thumb volar to the plane of the remaining digits. Intraosseous sutures through the metacarpal head and the dorsal strut of the index metacarpal are preferred to C-wires for fixation. Excellent retraction and meticulous attention to detail are required during this step, which often occurs 90 to 100 minutes after tourniquet inflation. The recessed index finger should lie in approximately 40 degrees of abduction and 120 degrees of pronation. Since the metacarpal head (now the thumb trapezium) is hyperextended, additional extension of the new thumb metacarpal is avoided. This maneuver is one of the most difficult during a pollicization procedure and requires careful retraction and assistance. The tourniquet is released for a 30- to 45-minute respite.

Intrinsic Rebalancing

Following recession and rotation of the index finger, the site of the backcut on the dorsum of the index finger is determined. This incision allows transposition of the ulnar skin to create a more aesthetic first web and a Y-V advancement of excess dorsal and radial skin from the palm (Fig. 41-25, *N* and *O*). It also provides wide exposure of the extensor mechanism for both intrinsic and extrinsic extensors reattachment. First the extrinsic extensors are shortened by approximately the same amount of metacarpal resection. The extensor digitorum is reattached to the base of the index proximal phalanx (now the thumb metacarpal) to simulate the abductor pollicis longus. Fixation on the ulnar side of the new metacarpal will help pronate the new thumb metacarpal. The extensor indicis proprius is interwoven into the extensor hood to act as the extensor pollicis longus. Next, the palmar interosseous (first PI) muscle is attached to the base of the index middle

phalanx (now the thumb proximal phalanx) to act as the thumb adductor pollicis. This muscle is often small and does not approach the new thumb with optimal mechanical advantage.

Occasionally, an excellent first PI and adductor pollicis are present with type IIIB thumbs. The first dorsal interosseous (first DI) (radial indicis, abductor indicis) muscle of the index finger becomes the new abductor pollicis brevis. If two muscle bellies are present, the deeper of the two is sutured to the radial base of the middle phalanx (now thumb proximal phalanx), and the other is then interwoven into the extensor mechanism distally at the level of the index PIP joint (now the thumb IP joint). Tension should be adjusted so that the thumb MP and IP joints are extended with the metacarpal in an abducted and pronated position. If the extrinsic flexors are not shortened, over time these will take up and provide more flexor tone. If they are shortened, the FDP is shortened by the same length as the metacarpal resection and done in zone III. The FDS tendon in this situation has been used as an immediate opponensplasty.⁹¹

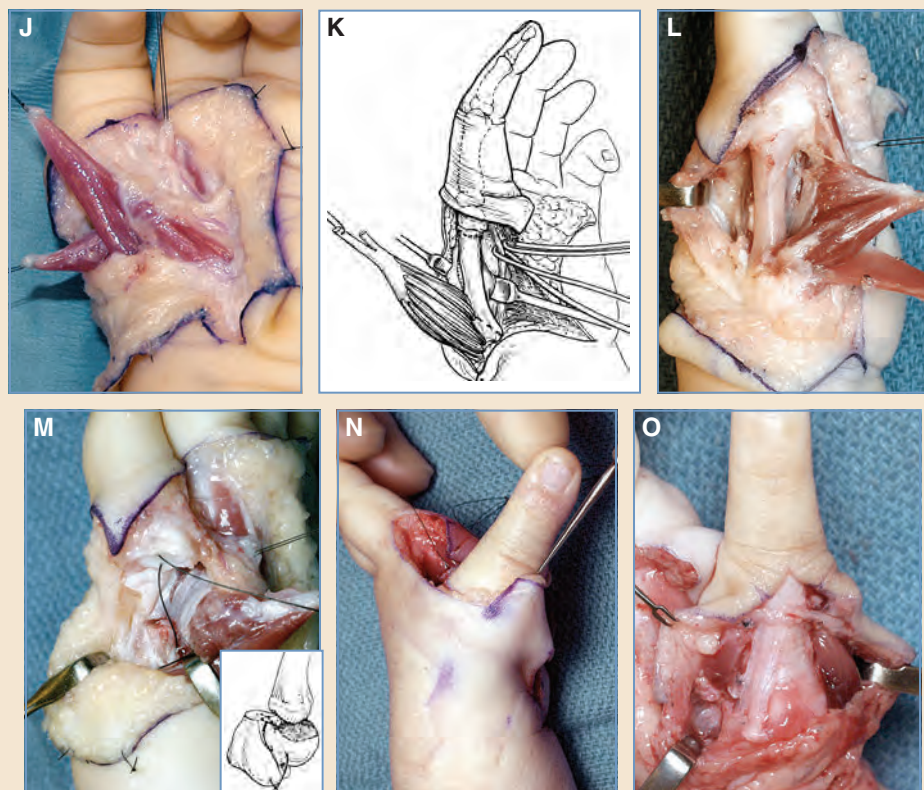


Fig. 43-25, cont'd **J**, The first dorsal interosseous (abductor indicis) muscle has been detached from its distal bone and extensor insertions. Two distinct muscles are often found. **K** and **L**, The muscles are attached to their periosteal origin, which has been elevated off the metacarpal. The distal osteotomy is through the epiphysis and the proximal cut leaves the dorsal cortex of the metacarpal base. **M**, The metacarpal head becomes the new thumb trapezium and is placed in a hyperextended position in front of the metacarpal base (*inset*). Interosseous suture fixation is preferred to C-wires, which can cause problems in young children. **N**, With the thumb in its new position, the available skin is then draped over the dorsal surface before the cut-back incision is made. This maneuver makes maximal use of all available tissue. **O**, The extrinsic tendons have been shortened and reattached, and the intrinsic muscles have been attached distally, either to bone or the extensor mechanism via the lateral bands.

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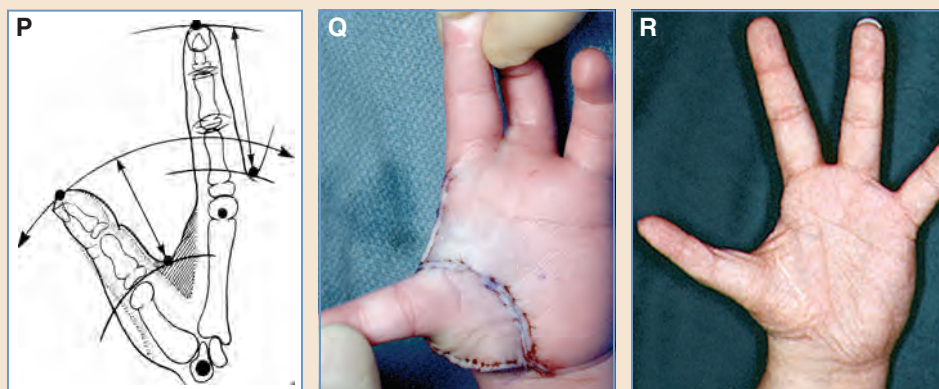


Fig. 43-25, cont'd **P**, The normal-appearing first web space is a gentle curve between the MP joint of the thumb and that of the long finger. Shaded area represents the area covered by the additional flaps rotated to create this web. **Q**, After closure, a broad web allowing maximal abduction should be present. **R**, The same hand is seen 21 years later.

Filletting the hypoplastic thumb remnant with removal of the bone, skin, and nail complex can subsequently be used for soft tissue augmentation of the new thenar region to assist in aesthetic improvement of the outcome.⁹⁴

Skin Closure

An eversion skin closure using both 5-0 and 6-0 catgut sutures starts at the base of the palmar incision. This closure helps to stabilize the position of the new thumb. The dorsal skin from the index has been advanced toward the long ray, so that an aesthetic thumb web extends from the MP joint of the new thumb and the MP joint of the long finger. Failure to do this will create an unnecessarily deep web space and give the appearance of a finger placed in the thumb position (Fig. 43-25, *P*). The flap advanced into the dorsum of the new thumb often contains an excess of subcutaneous fat. Debulking can be done, but only with great care to avoid injury to the venous drainage of the new thumb. No drains are required. After closure, a broad web allowing maximal abduction should be present (Fig. 43-25, *Q*). The patient is seen 21 years later (Fig. 43-25, *R*).

Dressing

Single-layer gauze impregnated with petrolatum is placed over the incisions and covered by a 2.0 to 3.0 cm thick layer of moistened cotton, which is molded to conform to the hand and new thumb. A bulky hand dressing is then applied, followed by application of a long arm cast well proximal to the elbow, which is flexed at 90 degrees. The thumb tip is left exposed to observe the circulation postoperatively, and the limb is elevated. The cast is removed 4 weeks postoperatively, and a thermoplastic thumb spica splint is fashioned with the thumb in abduction; this is to be worn at night for 2 months. During the day, unrestricted play is allowed. Many months may pass before active integration of the new thumb into hand function occurs. Buddy taping of the long and ring fingers is encouraged for children who persist with scissoring instead of thumb-long finger pinch or grasp.

Follow-up

These children require yearly follow-up to assess function and observe growth. As the first dorsal interosseous may be hypoplastic or absent in these children, secondary procedures are often required to achieve adequate palmar abduction. Transfer of the abductor digiti quinti is most frequently chosen. Other procedures that are occasionally required include extensor tendon shortening, tenolysis, scar revisions, debulking of dorsal fat, and dog-ear resection.

It is important to inform the parents that the pollicized index finger is not a thumb, but simulates a thumb. It will always be deficient in width, pulp, nail width, and joint motion. Manske and colleagues^{75,95} reviewed the function of pollicizations and found that the total active range of motion and grip strength was 50% and 21% of normal, respectively. Ninety-two percent of patients used the new thumb when manipulating large objects, whereas 77% did for small objects. Even though most patients used the thumb for performing tasks, it took them an average of 22% longer to do so. These results were adversely affected if the patient had an associated upper limb difference, such as radial dysplasia, a five-finger hand, or the mirror hand anomaly.

The most predictive preoperative variable is the condition of the index digit. Children with normal active and passive range of motion who have a normal component of intrinsic muscles and a normal distal radius and carpus will achieve much better results from a properly performed pollicization.

Treatment of a Stiff or Inadequate Index Finger

There are a number of clinical situations in which a less than adequate index digit is available for transfer into the thumb position. These include:

- Holt-Oram syndrome with a syndactylized index ray (Fig. 43-26)
- Index finger joined to the long finger with a complete simple syndactyly (that is, typical cleft hand, index-long finger syndactyly with absent or hypoplastic thumb)
- Stiff index finger associated with a complete or partial absence of the radius (Figs. 43-27 and 43-28; see Fig. 43-26)
- Stiff index ray with a fixed proximal interphalangeal joint flexion contracture, with or without a complete or partial absence of the radius (see Fig. 43-27)
- Mirror hand (ulnar dimelia), or the five-fingered hand

The clinical deficiencies of the index ray are obvious on physical examination; the major question is whether anything can be accomplished surgically to improve either the clinical function or appearance of the hand. The absence of flexion creases indicates deficient joint structures plus or minus inadequate extrinsic flexor or extensor myotendinous units. The presence of a complete or incomplete syndactyly means that there has been very little motion in utero. At surgery fibrous bands are present between the phalanges. Flexion contractures (camptodactyly) may be present in the index finger or all digits. These clinical situations do not present frequently but occur more often than one may anticipate. In our own combined experience, with more than 400 pollicizations performed over the past 29 years, one of these clinical scenarios is present at least 15% of the time.⁹⁰ There are no standard guidelines for treatment in the literature, and surgical recommendations are dependent on a given surgeon's experience. Most recommendations have been overwhelmingly conservative.*

Options for reconstruction include the following:

- Doing nothing
- Rotation-recession osteotomy of the index ray
- Formal pollicization of the index ray
- Pollicization of the fifth finger

*References 33, 75, 88, 90, 93, 95-106.

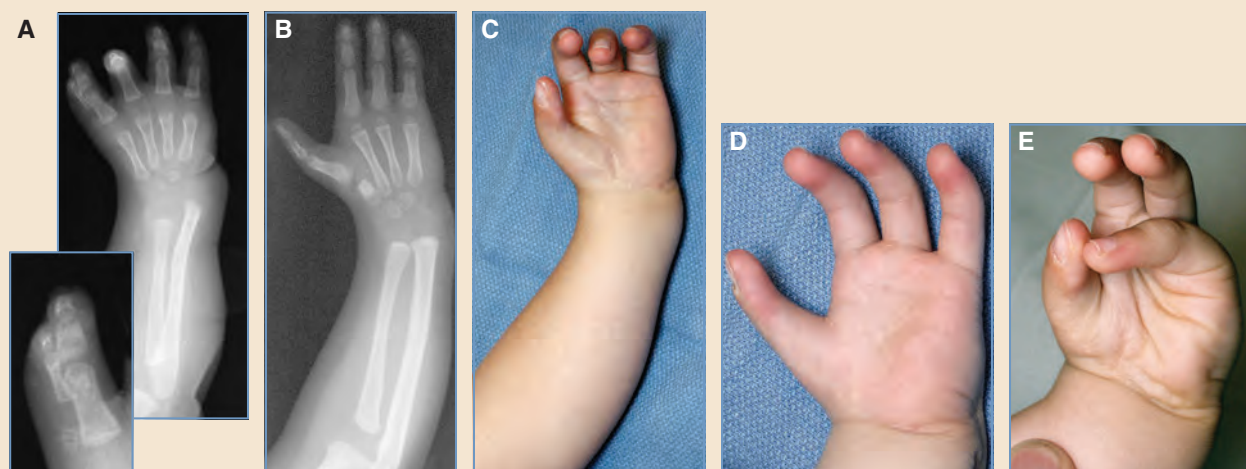


Fig. 43-26 Stiff index digit and radial dysplasia. **A**, This 5-year-old boy, who was born with an absent thumb and partial absence of the radius, underwent a centralization at 8 months of age and release of an index PIP camptodactyly at 1 year of age. **B** and **C**, Six months later, a rotational-recession osteotomy of the stiff index ray was performed, and **D**, the growth of the metacarpal head arrested 5 years later. **E**, Although this thumb has limited motion and slight excess in length, a very functional pinch has been maintained. The appearance is good. A strong index–first dorsal interosseous muscle, now the thumb abductor pollicis brevis, has stabilized the palmar abduction. The radiograph demonstrates the characteristic persistent radial deviation despite a good centralization with broadening of the distal ulna, which now looks more like a radius than an ulna.

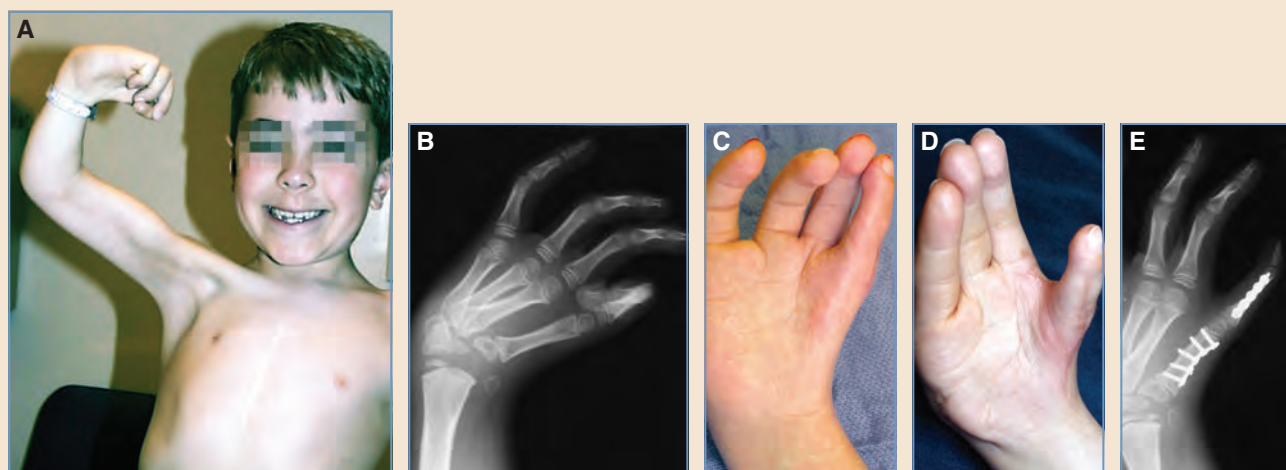


Fig. 43-27 Rotation-recession osteotomy versus pollicization. **A**, This proud 7-year-old flexes his pectoralis major muscle transfer for elbow flexion. The hand and carpus had been centralized earlier, and his hand function was notable for good flexion and extension of the ring and fifth digits and, to a lesser extent, the long finger. The immobile index finger was scissored beneath the long finger with a 90-degree fixed flexion contracture of the PIP joint. **B**, This radiograph shows the characteristic widening of the distal ulna, which now looks like a radius several years after centralization. The index ray has good skeletal parts in a nonfunctional position. **C**, Following partial excision and arthrodesis in extension, the index finger was used as a post for the long finger. **D**, The incision for a rotation-recession osteotomy at the metacarpal level used the Y-V principle. **E**, The digit was shortened 8.0 mm and abducted to create a wider first web space.

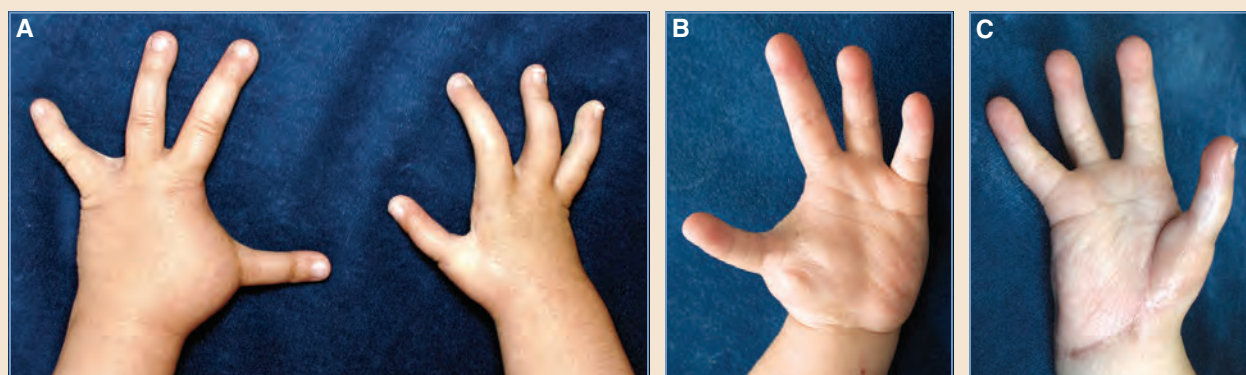


Fig. 43-28 Thumb position after pollicization. **A**, This child, seen 6 years after bilateral pollicizations, had a normal radius and index digit on the left and a short radius with a stiff index ray on the right hand. **B**, The left thumb, with excellent motor function, can be placed in more palmar abduction and extension. **C**, The stiffer right thumb is placed closer to the long finger so that tip-to-tip prehension is possible. The right thumb would be a functional liability if placed in the same position as the more mobile left thumb. Before pollicization on the right side, camptodactyly releases were performed at the PIP joint level of all four digits.

The possible combination of deformities with congenital hand differences is infinite, and there are many instances in which it is possible to pollicize an index digit after a number of previous procedures involving this ray have been completed. Most examples involve either a syndactylized index finger within a mitten hand or a typical cleft hand in which the index and long fingers are joined within a simple syndactyly. In either case, the syndactyly is released before formal repositioning of the index digit.

Determining what to do and how to do it can be made much easier by analysis of larger series of pollicizations. The quality of the result depends predominantly on the preoperative condition of the index ray. Optimal results are predictable following a well-executed operation on an index finger with normal active and passive range of motion and a full complement of median- and ulnar-innervated thenar intrinsic muscles. The radius is usually normal in these children. In the five conditions listed on p. 1329, the index and long digits have diminished motion, and the ring and small (fifth) digits are mobile and functional. In these hands the position of the pollicized index finger is crucial so that it will be well positioned for a pinch to the long finger and a grasp to the fifth finger. In most cases the new thumb will have diminished extrinsic motion, particularly in extension, because the muscles originating on the radial side of the forearm are either abnormal or absent. It is most important that this thumb be positioned for good opposition to the fifth finger, a posture that places it in less abduction than normal (see Fig. 43-28). If placed in too much palmar abduction and extension, this immobile post will be caught on objects or within pockets.

One alternative rarely chosen in these patients is the rotation-recession osteotomy in lieu of a formal pollicization. We have preferred this procedure in teenagers and adults with a stiff, flexed index finger (see Fig. 43-27), those with scarring from previous procedures, mirror hands, a five-fingered hand, and a very stiff index ray associated with a radial club hand. The principles of treatment are the same, but the ray is not shortened to the full extent of a formal pollicization. What intrinsic muscles are present are reattached and the extrinsic flexors and extensor tendons are shortened as appropriate. The position of the new thumb must be very carefully chosen for optimal pinch. Many of radial club hand patients are born with a camptodactyly involving all proximal interphalangeal joints, which become progressively more severe in the most radial index

and long digits (see Figs. 43-27 and 43-28). In these contracted digits we also perform full releases with Z-plasties or full-thickness skin grafts if needed and then consider a rotation-recession osteotomy instead of a classic pollicization.

When assessing the outcomes of pollicization, one needs to consider whether there is an isolated thumb anomaly or if there is an associated forearm condition. In studies that have looked at the outcomes of pollicization, those children with an isolated thumb anomaly performed better after pollicization compared with those with concomitant radial dysplasia. The total active range of motion (TAM) of pollicized index fingers in children with isolated thumb anomalies is reduced (89 to 146 degrees) when compared with age-matched children with normal thumbs (185 degrees). If there is associated radial dysplasia, there is even a further reduction in the TAM of those pollicized digits.^{95,107-109} Strength, as measured by pinch and grip strengths, is also reported to be significantly less than age-matched normals in children following pollicization. When comparing strength in isolated thumb anomalies these fare better than those with associated radial dysplasia.^{95,107-109}

Refinements in Index Pollicization

Most surgical procedures represent a dynamic process and are amenable to alteration and refinement and pollicization is no exception. Although the very specialized elements of the thumb such as the basal saddle joint, the asymmetric condylar surface of the proximal phalanx and the contoured pulp surface and the broad distal phalanx are impossible to replicate, a close replica

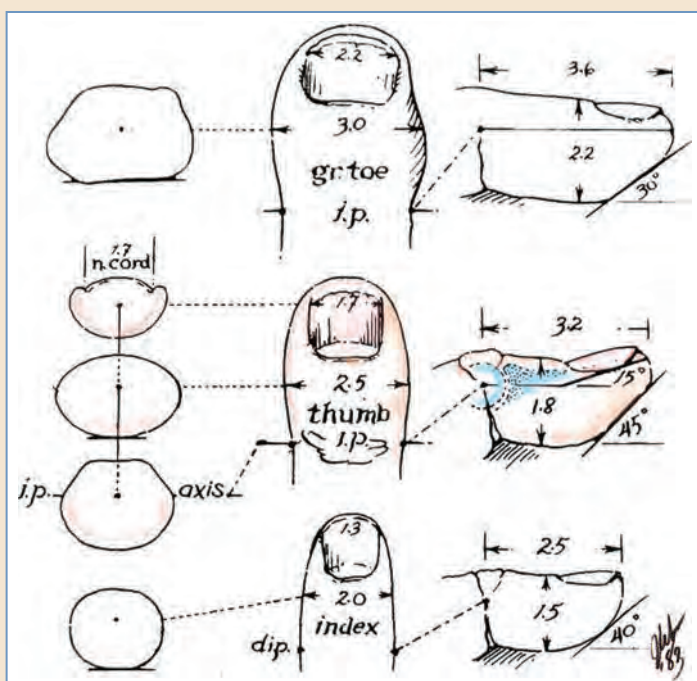


Fig. 43-29 The highly specialized pulp surface of the thumb with the great toe and index finger is compared. The contoured thumb with its slightly upgraded contact surface cannot be concisely replicated by these close substitutes.

is possible (Fig. 43-29). Working over a span of almost 80 years, the senior author (J.U.) and his mentor (J.W. Littler) have studied their craft critically.^{110,111}

Incisions

With the basic rotation and recession of the index ray into the thumb position, all incision designs involve a Y-V manipulation of skin, and this operation is no exception. Initially, Littler placed the V at the radial base of the index ray and made a small cut-back incision for the inset. The result was a new thumb in good position with imperfect contour within the first web space¹¹² (Fig. 43-30). The contour between the new thumb and long digit was not a gentle one, which flowed from the MP joint of the new thumb (previously the index PIP joint) and the MP joint of the long digit.

Two refinements have subsequently been made. The first places the palmar incision in what would be a normal thenar flexion crease (see Fig. 43-25) in a more central position and the second has been to delay the cut-back incision until the index has been placed into its new position. At that time the skin over the dorsal surface can be draped over the new thumb and a more precise cut can be made on the dorsal surface. This is usually over the mid-dorsal surface up to the level of the MP joint (previously the index PIP) and allows for easier rotation of the tissue on the ulnar

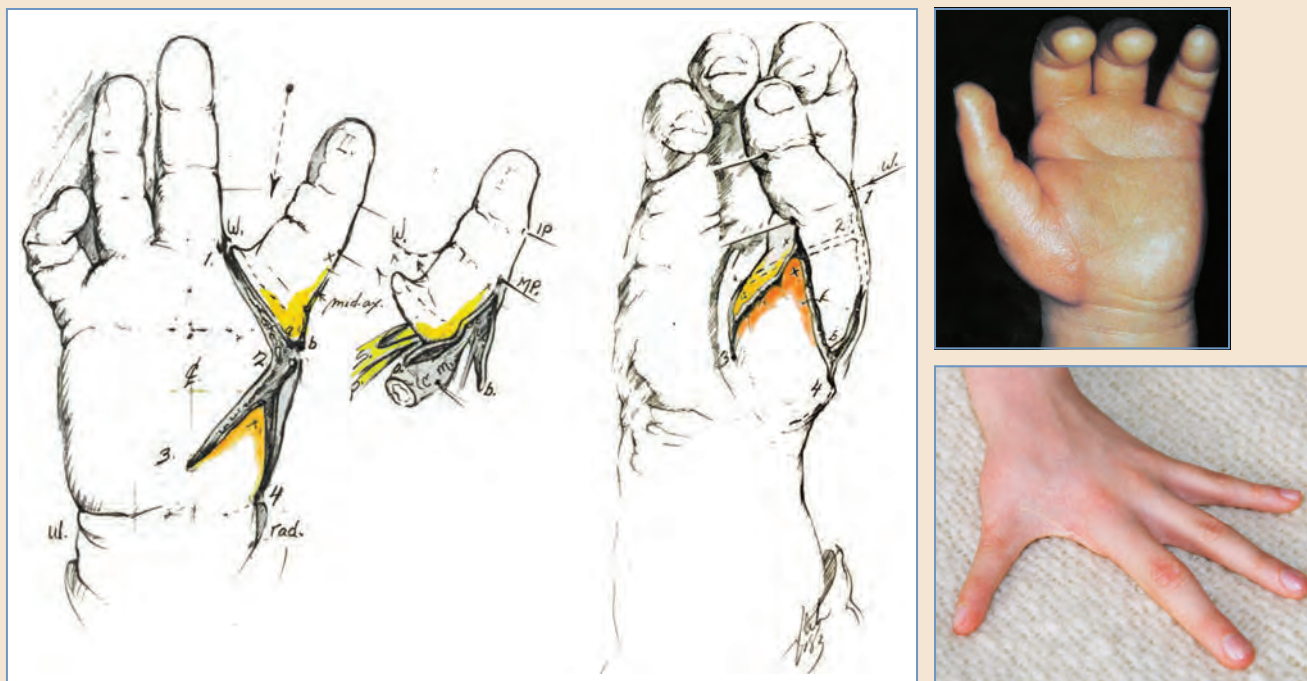


Fig. 43-30 Original incision design. Littler's original design placed a cut-back incision on the radial base of the index finger, which allowed the inset of a V-shaped flap of thenar skin. This resulted in a V-shaped cleft between the thumb and long digit and a thenar flexion crease on the radial side of the hand. The refinements discussed have created a more natural-appearing web, which extends from the MP joint of the new thumb to the MP joint of the long digit.

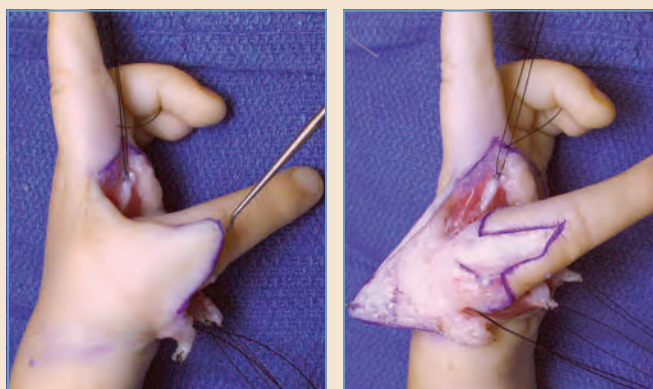


Fig. 43-31 Cut-back incision. Once the rotation-recession of the index ray has been completed, the dorsal tissue can be draped over the thumb in its new position. Its apex is marked and an incision made over the proximal phalanx of the index digit, which is now the new thumb metacarpal. In this case, the most appropriate incision was in the middle. However, it may also fall along the radial or ulnar surface dependent upon the soft tissue available. The skin on the ulnar surface is then available to be advanced into the more contoured first web space.

side of the index finger into the new web space (Fig. 43-31). The delay in making this strategic incision enables the surgeon to make maximal use of all soft tissue available.

Carpometacarpal Joint

The normal thumb carpometacarpal (CMC) joint represents an absolute marvel of engineering: it permits motion in four directions (abduction, adduction, flexion, and extension), plus limited rotation in pronation and supination. In a hyperextended position, the index MP joint permits only flexion and extension. The original pollicization design simply shortened the metacarpal and placed the metacarpal head at its base. With the head in an extended position, the new thumb would not hyperextend in its new position. The refinement has been to make certain the growth plate has been obliterated. Growth of the metacarpal head is not desired, since the transferred metacarpal head is close to the size of a normal trapezium. With the joint in a hyperextended position, the index metacarpal head (now the new thumb trapezium) is placed anterior to the base of the index metacarpal. The previously described osteotomy is made at the base and the metacarpal head sutured into its new position. The metacarpal head is sutured into its new position and the use of pins is avoided (Fig. 43-32). (C-wires in children are often the source of problems.)

Extrinsic Tendons

The refinement in technique has been to use the index extensor digitorum communis as a new abductor pollicis longus, which is inserted onto the ulnar base of the new thumb metacarpal (previously the index proximal phalanx). In this new position, it will effectively extend the new thumb metacarpal and pronate as much as the new CMC joint will allow¹¹² (Fig. 43-33).

Intrinsic Muscle Rebalancing

The rebalancing of the intrinsic muscles is critical for the stabilization of the new ray in the thumb position. There is great variation in the size and quality of these muscles and their dissection and reinsertion requires experience. We prefer to insert the first dorsal interosseous (first DI) (also called *the abductor indicis*) muscle directly into bone, because this single insertion gives it a direct

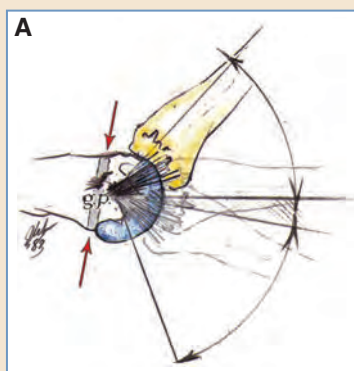


Fig. 43-32 CMC joint. **A**, The distal osteotomy of the index metacarpal is made through the growth plate at the level of the collateral ligaments. **B**, All epiphyseal cartilage is removed down to the level of bleeding cancellous bone. To avoid hyperextension of the new thumb, this joint is then placed in hyperextension in its new position. **C**, An oblique osteotomy is made at the base of the index metacarpal and the palmar cortical surface is removed. The recessed (shortened) and rotated metacarpal head is then inset into this new position and secured with an interosseous suture.

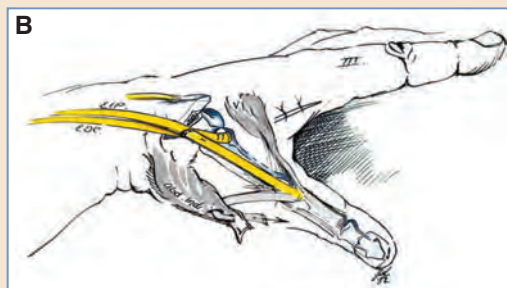
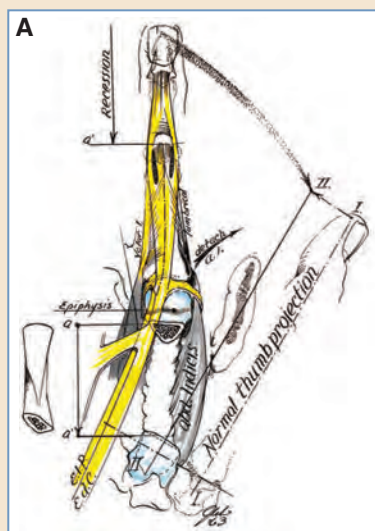
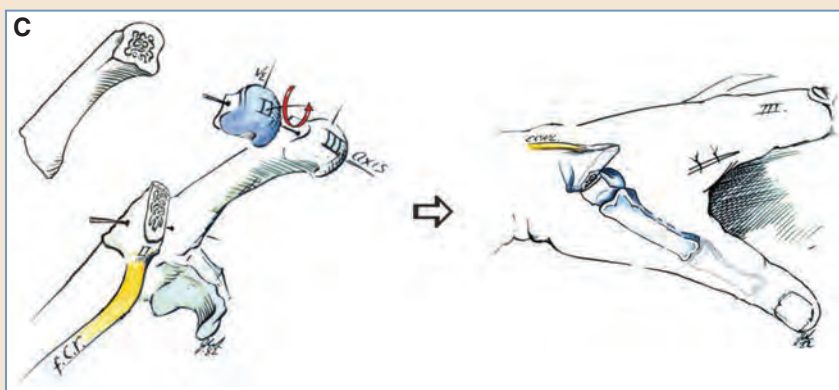


Fig. 43-33 Extrinsic extensor tendons. The two extrinsic tendons to the index digit, which must be rebalanced. **A**, A Littler drawing shows the tendons from the dorsal surface. In normal positions, the extensor indicis proprius is ulnar to the extensor digitorum communis. The abductor indicis is the first dorsal interosseous muscle. The new thumb projection is indicated. **B**, In the new thumb the extensor indicis proprius, an independent muscle tendon unit, is shortened to become the new extensor pollicis longus. The refinement is that instead of being sacrificed, the extensor digitorum communis is shortened and sutured to the ulnar base of the new thumb to become the abductor pollicis longus. With its pull exerted on the ulnar surface of the new thumb, additional pronation may be achieved. (*Detach ai*, Detach abductor indicis or first dorsal interosseous muscle; *EDC*, extensor digitorum communis to index digit; *EIP*, extensor indicis proprius; *Epiphysis*, growth plate in metacarpal head; *I*, thumb ray; *II*, index ray; *III*, long metacarpal; *Volar i*, volar interosseous muscle.)

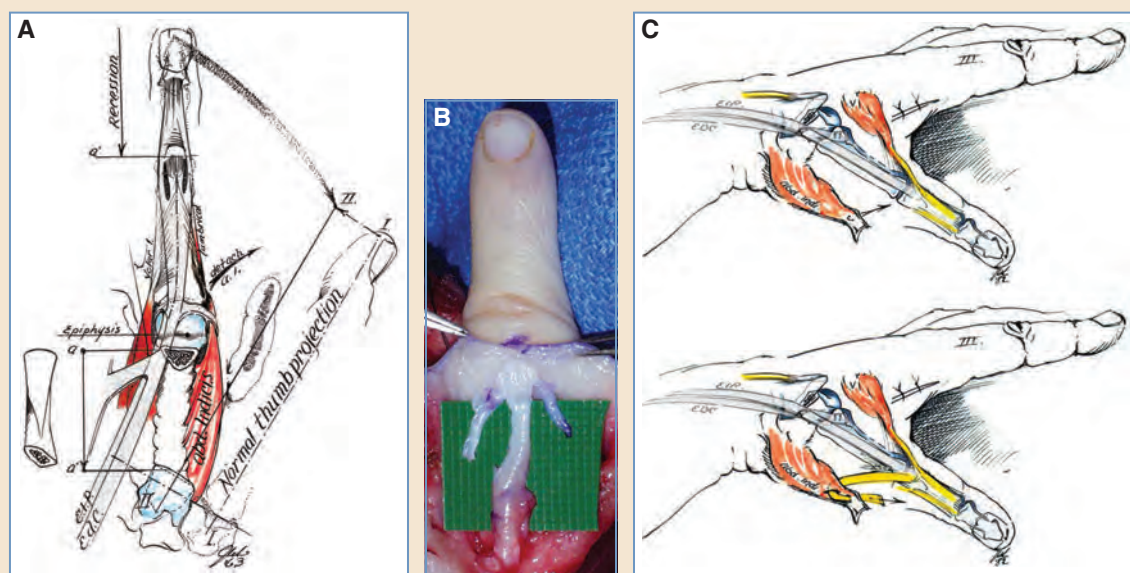


Fig. 43-34 Intrinsic rebalancing. **A**, In contrast to a normal thumb, which has three layers of intrinsic muscles, the index ray contains only two muscles that require reinsertion: the first dorsal interosseous (first DI) becomes the abductor pollicis brevis and the palmar interosseous, which becomes the adductor pollicis of the new thumb. **B**, The lateral bands have been prepared for intrinsic reinsertion on their side of the extensor mechanism. **C**, The volar interosseous has been attached to the ulnar lateral band of the extensor mechanism and a similar insertion for the larger abductor indicis. When two heads are present, dual skeletal and extensor insertions may be performed. (*Detach ai*, Detach abductor indicis or first dorsal interosseous muscle; *EDC*, extensor digitorum communis to index digit; *EIP*, extensor indicis proprius; *Epiphysis*, growth plate in metacarpal head; *I*, thumb ray; *II*, index ray; *III*, long metacarpal; *Volar i*, volar interosseous muscle.)

pull with all power concentrated at one point. In those with a normal index ray, this muscle often has two portions and the larger is preferably attached to bone and the smaller interwoven into the extensor mechanism. The older interweave of the lateral bands into the distal portion of these muscles is secure and often more stable than direct tendon-to-tendon sutures (Fig. 43-34).

In individuals with a stiff index ray, often associated with a complete or partial absence of the radius, the first DI is small and often insufficient to hold the new thumb in palmar abduction. Secondary tendon and/or muscle transfer is needed to provide this function. The palmar interosseous muscle is always much smaller—and equally as variable as the first DI. After the rotation and recession of the index ray, this muscle becomes the adductor pollicis of the new thumb. It may be inserted into either the lateral band or directly onto the ulnar side of the proximal phalanx or MP joint. The major problems with this often diminutive muscle is that it may cause too much adduction of the new ray. For this reason, we may provide additional length with a lateral band interweave or just discard the reinsertion if the muscle is small and has minimal excursion.

The final refinement here is to adjust the proximal origin of the first DI. When space is being cleared at the metacarpal base for the reception of the metacarpal head, the muscle origin and its periosteal attachments are often elevated. This maneuver provides an opportunity to adjust the tension of this muscle at its proximal origin; a move that occurs before the final skin closure.

Suturing

Comparison of preoperative and postoperative plaster molds of these patients up to 10 to 20 years later often highlights the dramatic appearance of a “railroad track” incision made with 5-0 sutures during the first few years of life. For this reason, we prefer to close these wounds in layers using 5-0 or 6-0 absorbable sutures for the deep layers and approximation of the epidermal surface with an intracuticular technique. Stitch marks are always more conspicuous in dorsal hair-bearing skin than in palmar glabrous skin.

Adipofascial Flap

In children with type IIIB or type IV thumb hypoplasia, the extra skin, fat, and/or pulp tissue does not necessarily need to be discarded. Much of this tissue can be saved as a vascular island and placed subcutaneously at the radial base of the new thumb to produce a simile of a thenar eminence.⁹³ This augmentation is not with a functional muscle but does create a significant improvement in appearance (Fig. 43-35).

Metacarpal Arch

Anterior placement of the metacarpal head during pollicization helps to simulate the normal concavity of the proximal palm and the transverse metacarpal arch (Fig. 43-36). It also allows easier fixation of the metacarpal head in permanent extension.

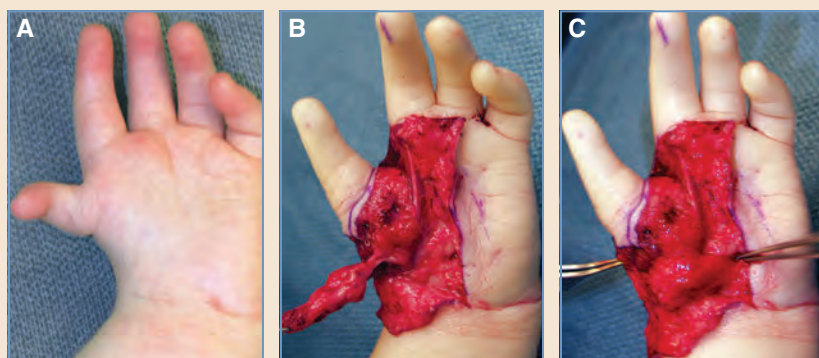


Fig. 43-35 Adipofascial flap. **A**, Following a fillet of skin and skeletal components, the remaining soft tissue of this type IIIB thumb has been preserved on **B**, a vascular pedicle and **C**, placed at the base of the new thumb. This additional bulk in a strategic area improves appearance.

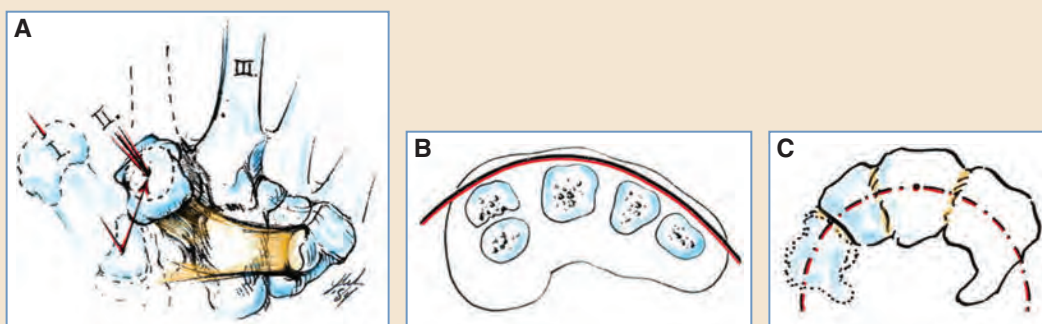


Fig. 43-36 Carpal arch. Anterior repositioning of the index metacarpal head. **A**, Coronal view; **B**, axial view. This helps to simulate the normal concavity of the proximal palm. **C**, The trapezium is outlined in dotted form in a normal carpal arch (axial view). (*I*, Thumb metacarpal; *II*, index metacarpal; *III*, long metacarpal.)

Other Revisions

Both function and appearance can be improved with secondary revision. The contour and breadth of the pulp can be augmented with composite glabrous pulp grafts harvested from the lateral side of the great toe (if present). Grafts with a width of up to 12 to 13 mm can be harvested with primary donor site closure in older children.

Lack of palmar abduction (opposition) can be improved with tendon or muscle transfers. Our preference for restoration of palmar abduction (opposition) has been the flexor digitorum superficialis passed around the flexor carpi ulnaris. The abductor digiti quinti muscle (Huber) transfer has also been used as our second choice. The major modification we have made with this transfer has been to include glabrous skin as a myocutaneous flap. The incision is the same along the ulnar border of the hand, and passage of the muscle through a subcutaneous tunnel is avoided, because a straight incision is made along the radial side of the thumb for direct inset. The complex relationship between the abductor and flexor pollicis muscles must be respected, since it is often wise to include both in the transfer. The opponens pollicis muscle can usually be left undisturbed at the base of the wound. This transfer has been used more frequently for type IIIA hypoplasias (Figs. 43-37 and 43-38).

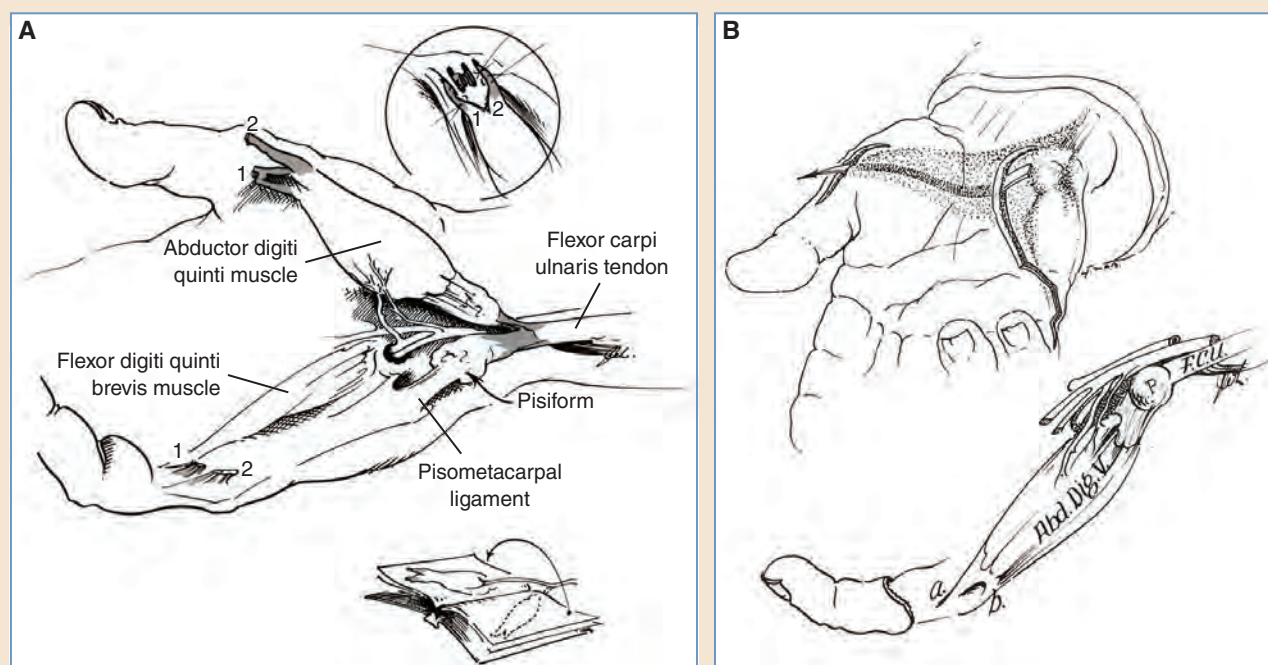


Fig. 43-37 Abductor digiti quinti muscle transfer (ADQM; Huber transfer). **A**, ADQM transfer for palmar abduction as muscle alone is isolated on its vascular origin within Guyon's canal. Additional length is achieved by split of the flexor carpi ulnaris. The skeletal (1) and extensor (2) insertions are anchored into the bone (2) and extensor (1) of the thumb. Transfer is likened to turning the page of a book. **B**, A subcutaneous path was initially preferred before the introduction of myocutaneous flaps. (*a* [and 1], *b* [and 2], Skeletal and tendinous insertions of the abductor digiti quinti muscle; *abd Dig V*, abductor digiti quinti muscle; *FCU*, flexor carpi ulnaris; *Flex Dig V*, flexor digiti quinti muscle; *P*, pisiform bone.)

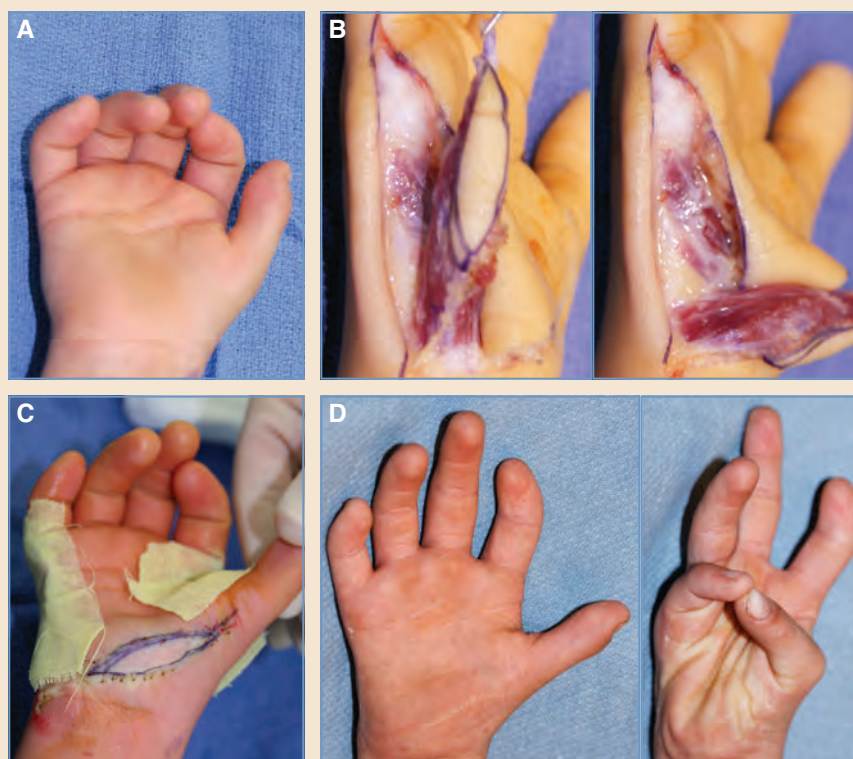


Fig. 43-38 Abductor digiti quinti myocutaneous flap/transfer. **A**, Appearance of a hypoplastic hand with type IIIA thumb hypoplasia with no functioning thenar intrinsic muscles. **B**, The abductor digiti quinti and flexor digiti quinti have been isolated together with a skin island and, still attached to the pisiform, the opponens digiti quinti remains intact. **C**, The resultant pull is directly across the palm. The skin island serves to further augment the base of the thumb. **D**, The muscle functions very well, and the color match and contour of the glabrous skin is ideal.

Central Deficiencies of the Hand With Thumb Abnormalities

There are two congenital hand differences that include central deficiencies within the hand and that were historically designated as the typical and atypical cleft hand. The latter is now called *symbrachydactyly*. Both conditions may have significant thumb malformation and are included as types of thumb hypoplasia.

TYPE VI CLEFT HAND: ECTRODACTYLY

Anatomy

Cleft hand is characterized by hypoplasia or aplasia of the central rays of the hand, thus forming a V-shaped or funnel-shaped cleft. All degrees of simple syndactyly of the first web are seen, resulting in moderate to severe deficiencies of the first web space. All degrees of hypoplasia extending to aplasia of the central two rays may exist. The ulnar two digits in the ring and fifth positions are commonly webbed with simple syndactylies. The thumb in the cleft hand anomaly is

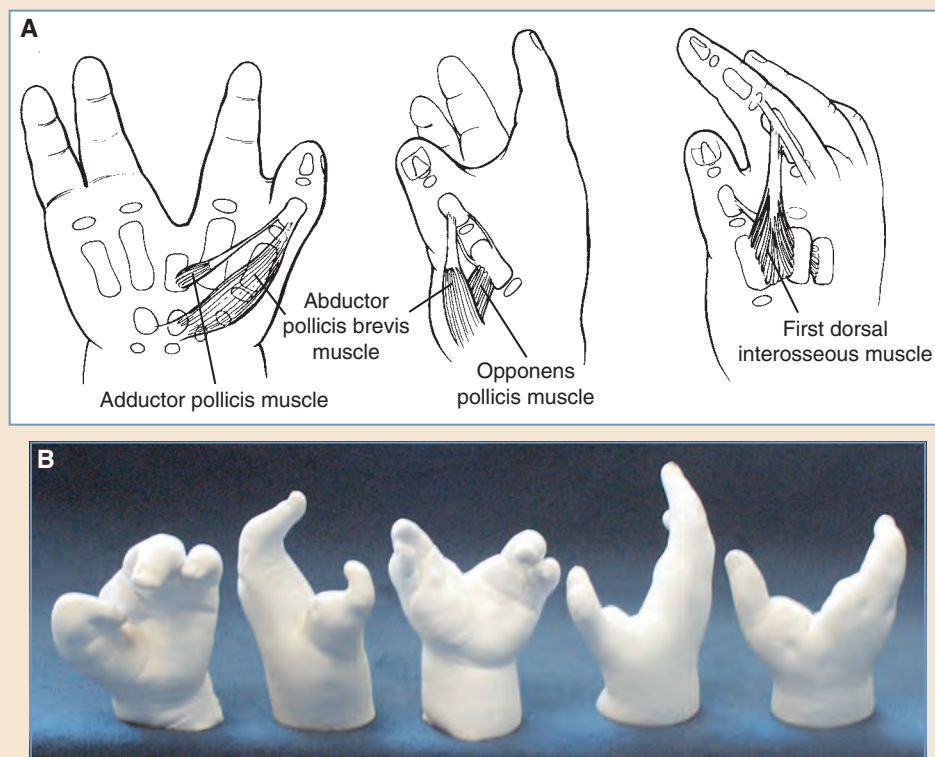


Fig. 43-39 Thumb hypoplasia and the cleft hand. **A**, Three layers of median-innervated intrinsic muscles are present in most typical cleft hands. Commonly, the index ray is joined to the thumb by a simple syndactyly. All degrees of absence of the long rays may be present. The first dorsal interosseous is present and tight, and the degree of adductor pollicis hypoplasia is proportional to the amount of the third metacarpal present. A very small adductor pollicis is illustrated. **B**, A sampling of molds shows the wide variation in the size and depth of the central clefting. The thumbs are smaller than normal, and the longer thumbs with flexion contractures are triphalangeal.

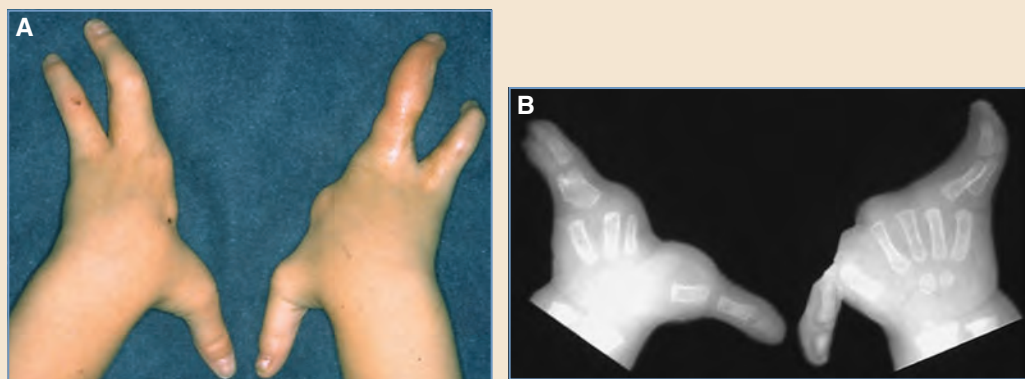


Fig. 43-40 Cleft hand. **A**, Clinical appearance and **B**, radiographs of both hands in this 4-year-old child show unstable thumbs at the MP joint level. Despite the wide central cleft and the absence of index and long digits, the child's function was remarkably good. Subsequent surgery consisted of ulnar collateral ligament reconstruction of both thumbs and a camptodactyly release on the right ring digit.

usually slightly smaller in size, with all components of the osteoarticular skeleton being present (Figs. 43-39 and 43-40). A Blauth type II thumb classification⁴¹ would be appropriate for a strict constructionist. The carpus and forearm bones are normal. The major deficiency of the thumb in this form of hypoplasia is that the ulnar-innervated intrinsic muscles are severely hypoplastic or absent. The median-innervated thenar intrinsic muscles are normal. In particular, the ulnar-innervated adductor pollicis is usually severely hypoplastic or absent, and the first dorsal interosseous muscle is moderately hypoplastic and contracted. Collateral ligament instability at the MP joint is common in most forms (see Fig. 43-40). The presence or absence of the third metacarpal is often an excellent indicator of the status of the adductor pollicis muscle. The extrinsic flexor and extensor myotendinous units to the thumb tend to be unaffected. A very important consideration in the correction of any cleft hand is the status of the long metacarpal and the adductor pollicis muscle (Fig. 43-41).

In another variation of cleft hand, thumb duplications at all levels may be observed. The more distal type I and II duplications are often associated with an absence of index phalanges with an intact metacarpal. Transversely oriented tubular bones at the distal metacarpal level are often seen with complete absence of the index (and long) digits. Superdigits are often seen in this variation.

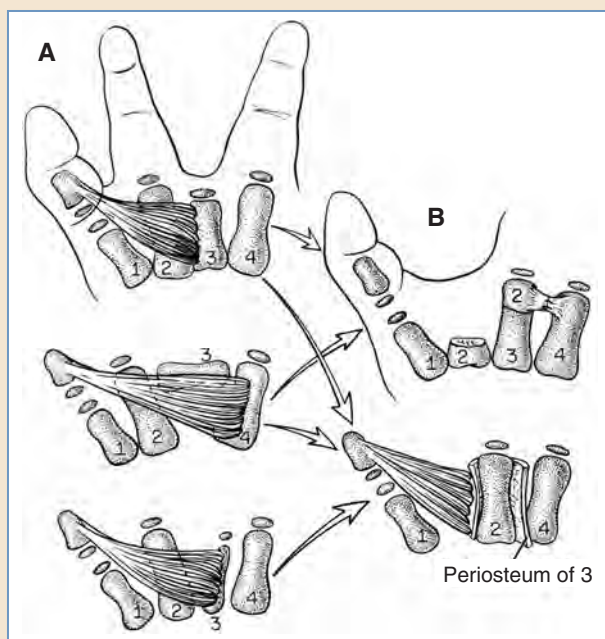


Fig. 43-41 The importance of the adductor pollicis muscle. **A**, A strong adductor muscle should be saved, because a weak pinch is a predominant feature of the cleft hand. Three common variations seen in cleft hands are seen on the left as a hypoplastic metacarpal with a strong muscle (*top*), a transverse phalanx with the adductor inserting into the next ulnar bone (*middle*), and a very hypoplastic metacarpal with a very deficient muscle (*bottom*). **B**, Two alternatives for treatment in these hands include the transposition of the distal index metacarpal on top of the existing long metacarpal (*top*) or a subperiosteal removal of a hypoplastic long metacarpal and transposition of the index into the periosteal sleeve (*bottom*). The transverse phalanges seen in the left center are excised completely. (1, Thumb metacarpal; 2, index metacarpal; 3, long metacarpal; 4, ring metacarpal.)

With more proximal type III, IV, and V duplications, the central (third) ray is often severely hypoplastic or absent. Triphalangeal duplicates may also be encountered with the proximal types.

Management

In treating a patient with a cleft hand anomaly, the following principles should be followed:

- Release of the first web syndactyly and contracture
- Ulnar transposition of the index finger to close the central cleft
- Construction of an intermetacarpal ligament between the second and fourth metacarpals
- Preservation of the adductor pollicis, if present, with its origin from the periosteum of the third metacarpal
- Transposition or rotation of the skin from the central cleft into the first web to cover the newly created web space (Fig. 43-42)
- Release of any syndactyly involving the two ulnar digits
- Treatment of the thumb duplication in a standard fashion (see Chapter 46)

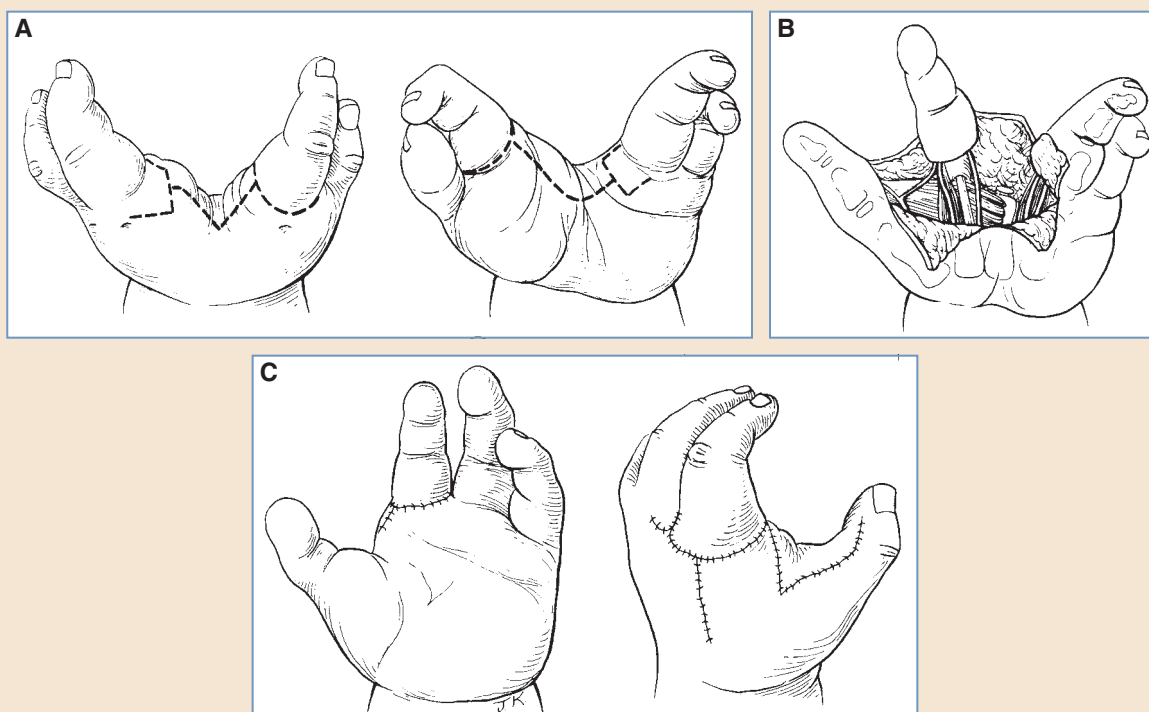


Fig. 43-42 Cleft hand correction incisions. **A**, Despite the degree of thumb hypoplasia, the approach to cleft hand remains consistent. Incision outlines on dorsal and palmar surfaces of the hand show a commissure flap on the radial side of the ring digit and equal division of tissue within the depth of the central cleft. All glabrous tissue remains palmar, and nonglabrous (hair-bearing) tissue remains dorsal. The major difference between our incision and the Snow-Littler incision is that we do not depend on a dorsally based flap to line the first web space. **B**, After flap elevation, there is wide exposure of all important neurovascular structures, the adductor pollicis (AddP), first dorsal interosseous (first DI) muscles, and all skeletal structures. There is easy access to all anatomy down to the CMC joint level. (This incision is the equivalent in the hand of the coronal incision used by craniofacial surgeons.) Complete exposure of all important structures is afforded by a direct incision. **C**, After transposition of the index ray, both flaps are moved into the first web space. Z-plasties or other transposition flaps can be used to improve contour.

Options

Several procedures have been described to accomplish the previously listed principles for the management of this condition.^{82,113-116} Flaps of the cleft skin based on either the volar or dorsal surface have been used. Both of these designs suffer from the same anatomic problem in that the skin flap from the cleft is a random-pattern flap, and the viability of the distal portion of the flap can be suspect. More recently, Tajima⁸⁴ has described closure of the newly created first web with a dorsal sliding flap. This design certainly has less risk associated with the vascularity of the skin flap.

Our preferred method of thumb correction uses simple incisions, much like those designed for a pollicization. An incision is made on the ulnar side of the index ray, which is then transposed as a vascular island to the long position. The metacarpal is either disarticulated and transposed at the CMC joint level, or, when a suitable portion of the third metacarpal base is present, osteotomy of the second metacarpal and transposition of this and osteosynthesis is then performed on top of the third metacarpal remnant. Dorsal and palmar flaps are raised and sutured directly to the skin flaps created by incision of the ulnar side of the cleft (see Fig. 43-42). Simple or complex Z-plasties can then be performed on either side of the thumb index web space as deemed necessary for contour improvement.

A number of autosomal dominant cleft hand and foot syndromes exist (Fig. 43-43). Genetic counseling may be required for those patients with bilateral cleft hand-foot anomalies, which are associated with an autosomal dominant pattern. In these patients a transverse failure of formation of the thumb may be present at the metacarpal or phalangeal level. Vascularized toe-to-thumb transfer using the distal portion of the great toe (if present) is preferred for thumb reconstruction when no phalangeal segments are present. Distraction lengthening is another alternative for lengthening. Motion in these thumbs is poor, sensation is good, and function is very gratifying, because the deformities are always bilateral. Secondary procedures are commonly needed for correction of intrinsic joint laxity or deviation.



Fig. 43-43 The thumb in a cleft hand. **A** and **B**, The left hand and radiograph of a young child with an unusual cleft hand shows a triphalangeal thumb, a duplication at the metacarpal level, a short index ray with no distal phalanges, and a transverse bone connecting the long and ring metacarpals at the MP joint level. **C**, The proposed reconstruction. In a first stage, the index ray (c) was transposed on top of the long metacarpal (f). Only the ulnar portion of the transverse bone (b) was saved because it articulated with the ring proximal phalanx.

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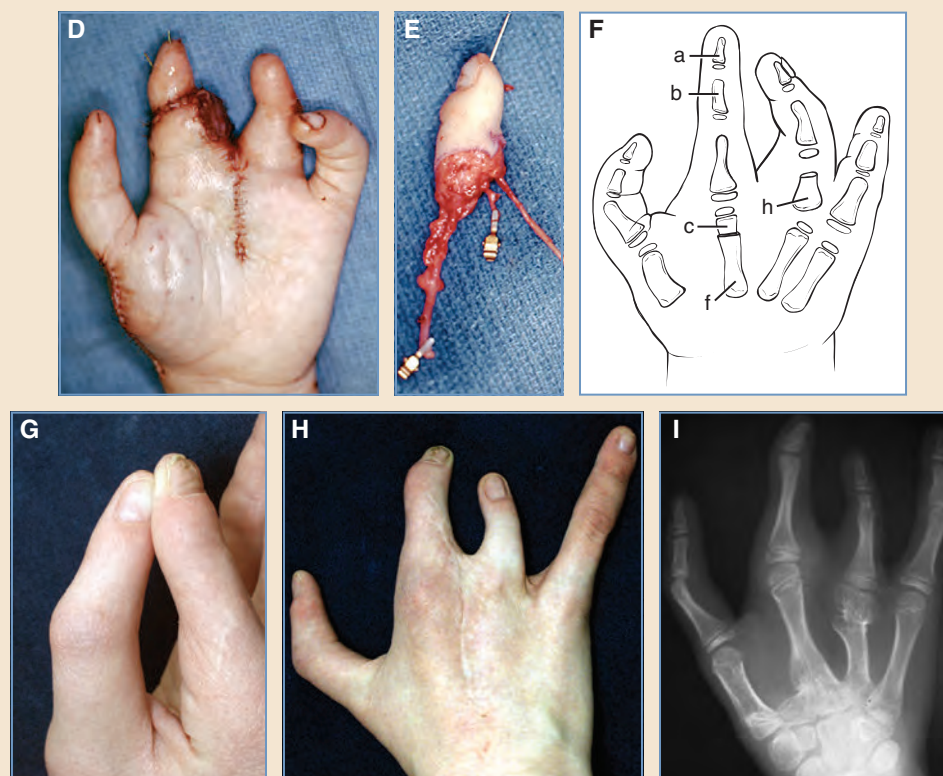


Fig. 43-43, cont'd D-E, At the second stage, the thumb duplication (*a, b*) was transferred on top of the index ray to make a longer and more complete digit. Nerves and tendons were all joined. G-I, The patient developed a very functional pinch between the long (and untouched) triphalangeal thumb and the index finger. Her hand and the radiograph are seen at skeletal maturity. The patient has become a very confident young woman and, in fact, has won regional New England piano competitions.

TYPE VII THUMB HYPOPLASIA: SYMBRACHYDACTYLY (ATYPICAL CLEFT HAND)

Anatomy

This form of longitudinal deficiency is always unilateral with varying degrees of hypoplasia of the central three rays of the hand. Nubbins with minute nail complexes may be present on the distal border of the palm representing the index, long, and ring fingers, but commonly there is complete absence of the central three digits, with varying degrees of hypoplasia of the central three metacarpals. All degrees of variation exist; no two hands are identical. The thumb in this condition is invariably smaller, with severe hypoplasia or aplasia of the adductor pollicis, but a metacarpal with two hypoplastic phalanges is usually present. Occasionally, a metacarpal and distal phalanx is present with a flail thumb at the MP joint level (Fig. 43-44). With an intact third metacarpal, a functional adductor pollicis muscle is often present. The median-innervated thenar intrinsic muscles are intact and usually small or normal. The thumb CMC joint is well segmented and mobile. The IP joint of the thumb may exhibit decreased passive and active motion as a result of weak extrinsic flexor and extensors. On exploration, the FPL tendon is often traced back to a hypoplastic or absent muscle belly in the distal forearm. In a severely hypoplastic thumb, there

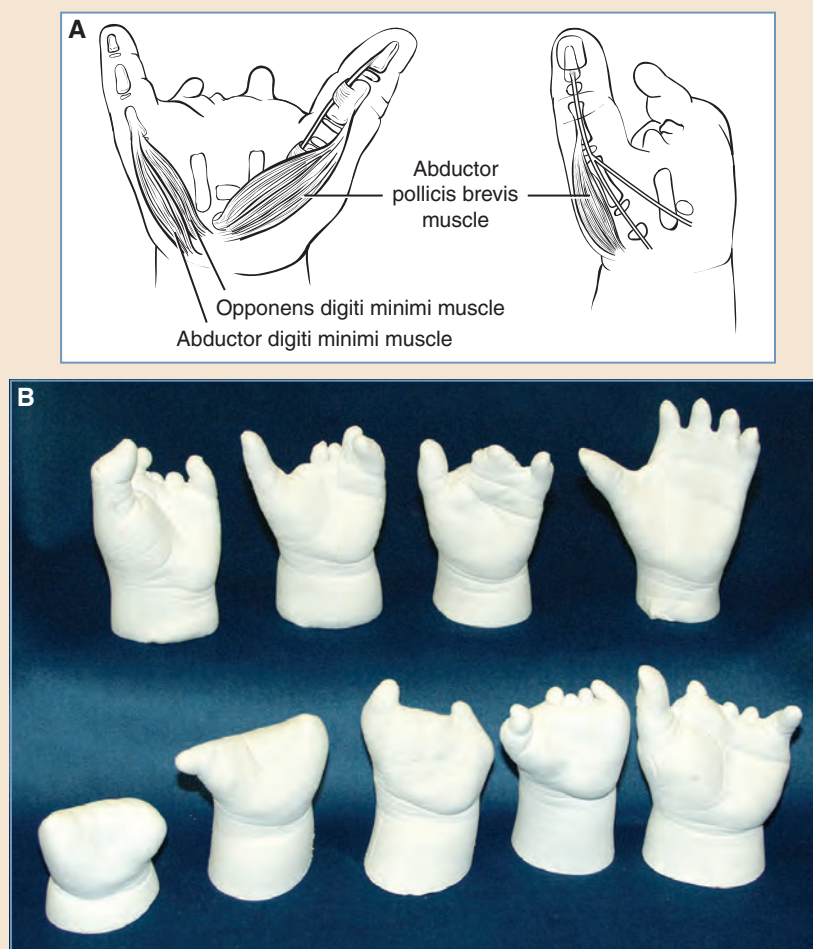


Fig. 43-44 Thumb hypoplasia and symbrachydactyly. **A**, The border thumb and fifth rays are the most complete in the hand. Thenar and hypothenar muscles are present and often small. Central digits are represented by hypoplastic nubbins. There are all degrees of metacarpal hypoplasia within the central three rays of the hand. Extrinsic flexors and extensors are present but abnormal. **B**, A very wide range of variation is present in these hands.

is significant limitation of MP motion, an absent IP flexion crease, and only a rudimentary FPL tendon. The fifth ray is usually the best in the hand, with intact intrinsic and extrinsic myotendinous units. Frequently this digit has a radial clinodactyly and completely unstable MP joint. The entire hand is hypoplastic. Radius and ulna are present and of equal length but may be small in comparison to the opposite limb.

Management

Observation of these children at play will provide insight into the need for any surgical reconstruction. Children with this congenital upper limb difference tend to be among the most functional of any, because virtually all can oppose and some can touch the thumb to the small finger. They all have the components of a basic hand: a mobile radial ray, a cleft (which acts as a web

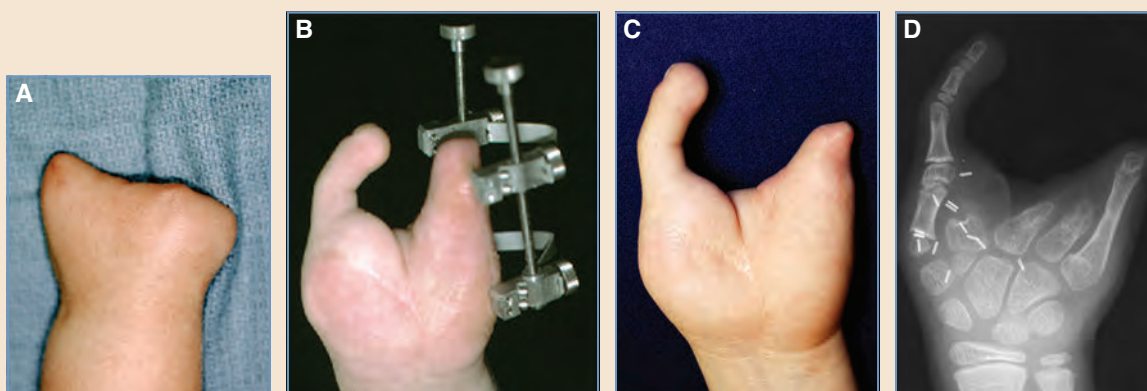


Fig. 43-45 Thumb in symbrachydactyly. **A** and **B**, The left hand of this child with Moebius syndrome was reconstructed with a second toe transfer at 18 months of age and is seen on the right when the patient was 5 years of age, during the distraction lengthening of the fifth metacarpal. The distraction was done in two stages: (1) osteotomy and application of distractor and (2) bone grafting. **C** and **D**, Ten years later, the second toe transfer continues to grow in comparison to the ulnar metacarpal post.

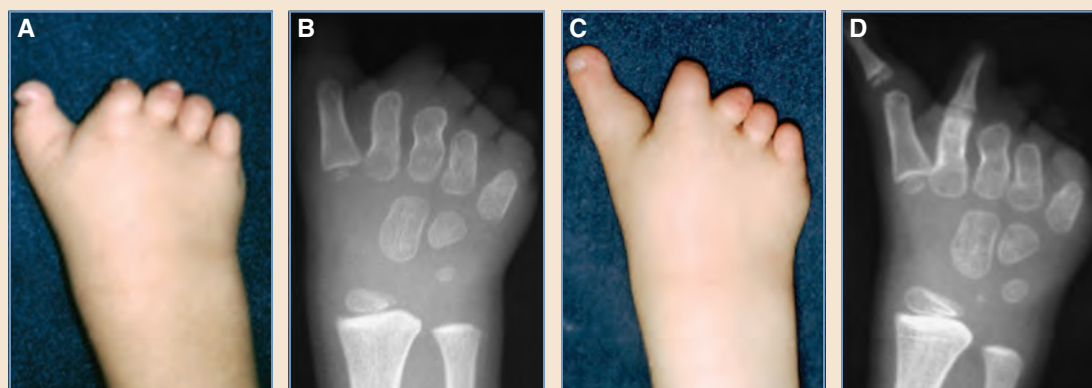


Fig. 43-46 Thumb in symbrachydactyly. **A** and **B**, The left hand and radiograph of a patient with generous soft tissue nubbins, which represent the thumb and digits. **C** and **D**, He was an ideal candidate for nonvascularized toe phalangeal transfer to the thumb and index rays. The first web space was also deepened and widened with local flaps. Despite the limited mobility, this reconstruction is very functional as a helper hand.

space) and an additional ray or post on the opposite side of the hand for pinching and grasping functions (Fig. 43-45). Consequently, most of these children do not require any operative intervention, and to provide it may be quite meddlesome.

Excision of the nonfunctional nubbins will improve the shape of the cleft and provide a deeper web space for grip. Some patients with this anomaly have a thumb deficient in length and unable to oppose to the highly mobile ulnar border digit. In these patients distraction lengthening can improve both function and appearance. These patients may also lack pulp-to-pulp pinch as a result of poor pronation of the thumb during pinch maneuvers. Rotational osteotomies at the metacarpal level will resolve this problem by placing the digits in a more favorable position. A severely hypoplastic thumb with an absent or diminutive proximal phalanx and a flail MP joint is best treated with a nonvascularized toe phalangeal transfer into the proximal phalangeal level.

Although motion is not restored, the thumb becomes a functional post. These transfers are more likely to grow if performed before the child is 1 year of age,^{117,118} although other studies have questioned the amount of actual long-term growth.¹¹⁹ Placement of a nonvascularized phalangeal transfer will provide bone stock for potential future distraction lengthening. Ablation of the small distal phalanx and second toe microvascular transfer is an option best performed between 2 and 4 years of age (Fig. 43-46).

The ulnar digit in symbrachydactyly will often have an unstable MP joint and radial clinodactyly as a result of its asymmetrical proximal phalanx. The flexor tendons are strong and the extensors present but unbalanced. Stabilization of the MP joint with chondrodesis, bone grafts, and transfer of nonvascularized toe phalanges have all been performed, with varying degrees of success. When positioned as a stable post, these fifth rays function quite well for hooking and balancing maneuvers.

TYPE VIII THUMB HYPOPLASIA: CONSTRICTION RING SYNDROME (AMNIOTIC BAND SEQUENCE)

Anatomy

Constriction ring syndrome (also called *amniotic band sequence* or *Streeter's dysplasia*) is a condition that can affect any one or all of the limbs and, uncommonly, the face. As defined by Patterson,¹²⁰ limb involvement can result in any of the following deformities^{82,120}:

- Simple constriction rings, which may be partial or circumferential
- Constriction rings with distal deformity, with or without concomitant lymphedema
- Acrosyndactyly (distal fusion, fenestrated syndactyly)
- Amputation

Hypoplasia of the thumb is seen in this condition when there is a deficiency in the length of the thumb, which may have a transverse amputation at any level (Fig. 43-47). Occasionally, the existing skeletal and soft tissue components of the first ray may be hypoplastic. The hallmark of the amniotic band sequence is that the anatomy proximal to the level of amputation or band is normal. Either superficial or deep constriction rings (annular bands) can be associated with hypoplasia or lymphedema of the distal segment of the digit with hypoplastic nail remnants and slender, truncated phalanges present. Acrosyndactyly usually involves the central three rays of the hand but can also involve the thumb and fifth finger in a large syndactylized complex. Amputation of the thumb in this condition is the major cause of partial aplasia of the thumb and, as previously mentioned, can occur at any position along its length. When considering the management of the level of amputation, it is best to do this at one of three levels: distal to the IP joint, the proximal phalanx, or the metacarpal (see Fig. 43-47). Motion of the IP joint is usually severely affected, even with amputations or deep constriction rings distal to it.⁸²

Management

The following principles apply to the management of these thumbs:

- Immediate treatment of emergency conditions such as vascular compromise and progressive lymphedema
- Early liberation of the thumb ray from an acrosyndactylous complex
- Release of adduction contractures within the first web space when present

After release of the first ray, the functional requirements of the patient must be accurately determined. Several options are available, and the management of each patient should be carefully individualized. In individuals with distal amputations at the phalangeal level, no operative intervention may be the best course of action.

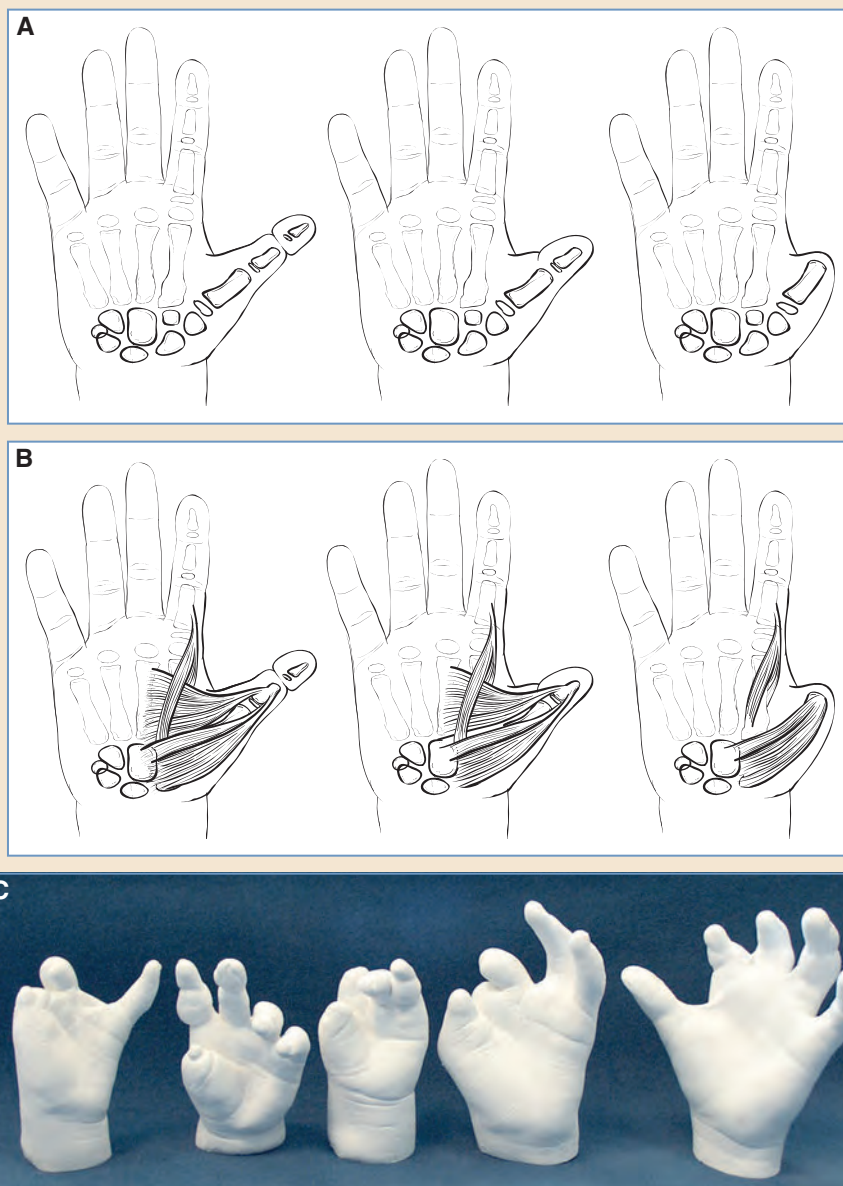


Fig. 43-47 Type IX thumb hypoplasia: constriction ring syndrome (CRS). **A**, The thumb in such hands may have a shallow or deep constriction ring or a congenital amputation at any level along the skeletal ray. The phalanges at the level of loss are characteristically narrow and taper to the distal stump. **B**, The intrinsic muscles are all present up to the level of the amputation. The proximal anatomy in this condition is normal. **C**, The thumb is absent on the left and shows increasing length in these molds. No two hands are identical; there is great variation in the depth and location of constriction rings along the digits.

Options available for lengthening the deficient thumb ray in this condition include distraction lengthening, nonvascularized toe phalangeal transfer, and composite vascularized toe-to-thumb transfer or local partial ray transfer.

Distraction lengthening^{121,122} is best performed at the metacarpal level if satisfactory bone stock is present. The metacarpal can be easily but slowly lengthened up to 100% of its length as a two-stage procedure, which includes application of a distractor and an osteotomy, followed by bone grafting within the intercalated gap. Lengthening of deficient, terminal, and often-narrowed phalanges in this condition is not as predictable. The resulting digits are often stiff and thin. Distal nonunions and exposures are common complications. Second toe composite transfers are preferred to distraction at the phalangeal level (Fig. 43-48).

Transfer of nonvascularized toe phalanges^{117,118,123} is an excellent way to provide length and growth potential to an empty, redundant soft tissue envelope occasionally seen distal to the level of amputation in this condition. Extraperiosteal harvest of the third or fourth toe proximal pha-

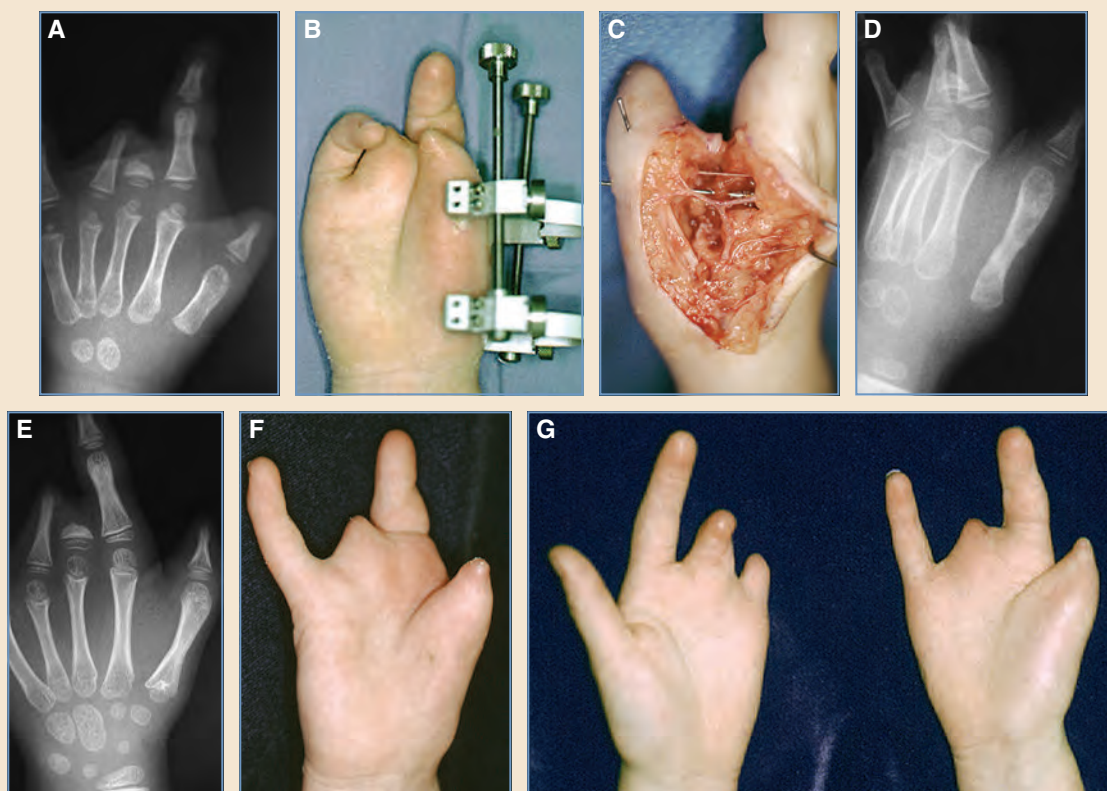


Fig. 43-48 Constriction ring syndrome treated with distraction lengthening. **A**, Radiograph of a young girl with a congenital amputation at the thumb interphalangeal joint level with a tight first web space. Note the tapered appearance of the proximal phalanx. **B**, A distraction apparatus is pulling each end of the metacarpal away from the osteotomy site at the middle diaphyseal level. One clockwise turn of the screw equaled a 1.0 mm gain. **C**, The new bone regenerate (*callostasis*) advances toward the midportion of the gap created. The stretching injury to soft tissue is impressive. All vessels, nerves, and tendons are intact. **D**, The thumb metacarpal is now as long as the index. **E**, Five years later, both metacarpals are growing, but the index is now longer. **F**, The first web space was deepened to increase thumb mobility and increase its span. **G**, Although shorter than normal, this thumb has functioned very well on her dominant hand.

lanx and transfer will result in survival and growth of the phalanx in 90% of patients younger than 2 years of age (Fig. 43-49). Our own experience has not been as successful: only 70% of these phalanges transferred early in life have retained normal growth potential. Survival and growth will occur in patients older than 2 years of age, but at a less predictable rate. The second toe is spared so that the entire toe is available for vascularized composite transfer if required. Secondary distraction lengthening of the transferred phalanx can be performed if further length is required.

In patients with a congenital amputation through the proximal phalanx or metacarpal, composite microvascular transfer of a second toe has been our procedure of choice. The great toe can be transferred as either a complete or as a modified unit,¹²⁴ but this option has been reserved for uncorrected thumb deformities that present after childhood. Because of the normal anatomy proximal to the level of amputation or band and an intact CMC joint, an excellent functional and aesthetic result of the “toe” can be obtained. The presence of functional thenar intrinsics will also enhance the result (Figs. 43-50 and 43-51).

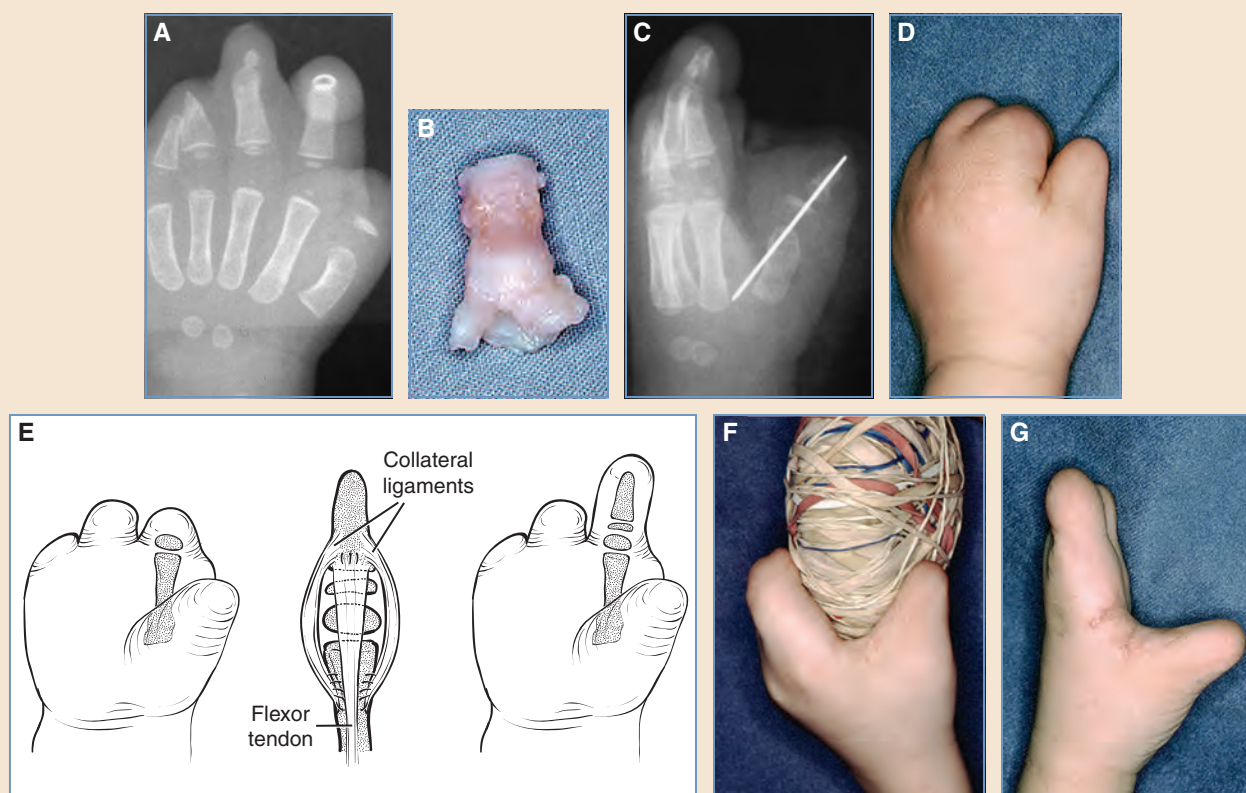


Fig. 43-49 Thumb with constriction ring syndrome treated with a toe phalanx. **A-D**, In this child with CRS acrosyndactyly, the fibrous connections holding all fingertips were released in the newborn nursery. Her thumb ends at the metaphysis of the proximal phalanx and the index finger at the middle phalanx. Nonvascularized toe phalanges from the third and fourth toes were transferred on top of the index finger and thumb. **E**, The periosteum is intact, and the collateral ligaments and volar plate are attached to the skeletal part at the amputation stump. Joint motion was preserved in this child. **F** and **G**, A four-flap Z-plasty deepened the first web space and improved her grasping ability. She is now 20 years of age and has had normal growth of the transferred phalanges. No further reconstruction has been performed.

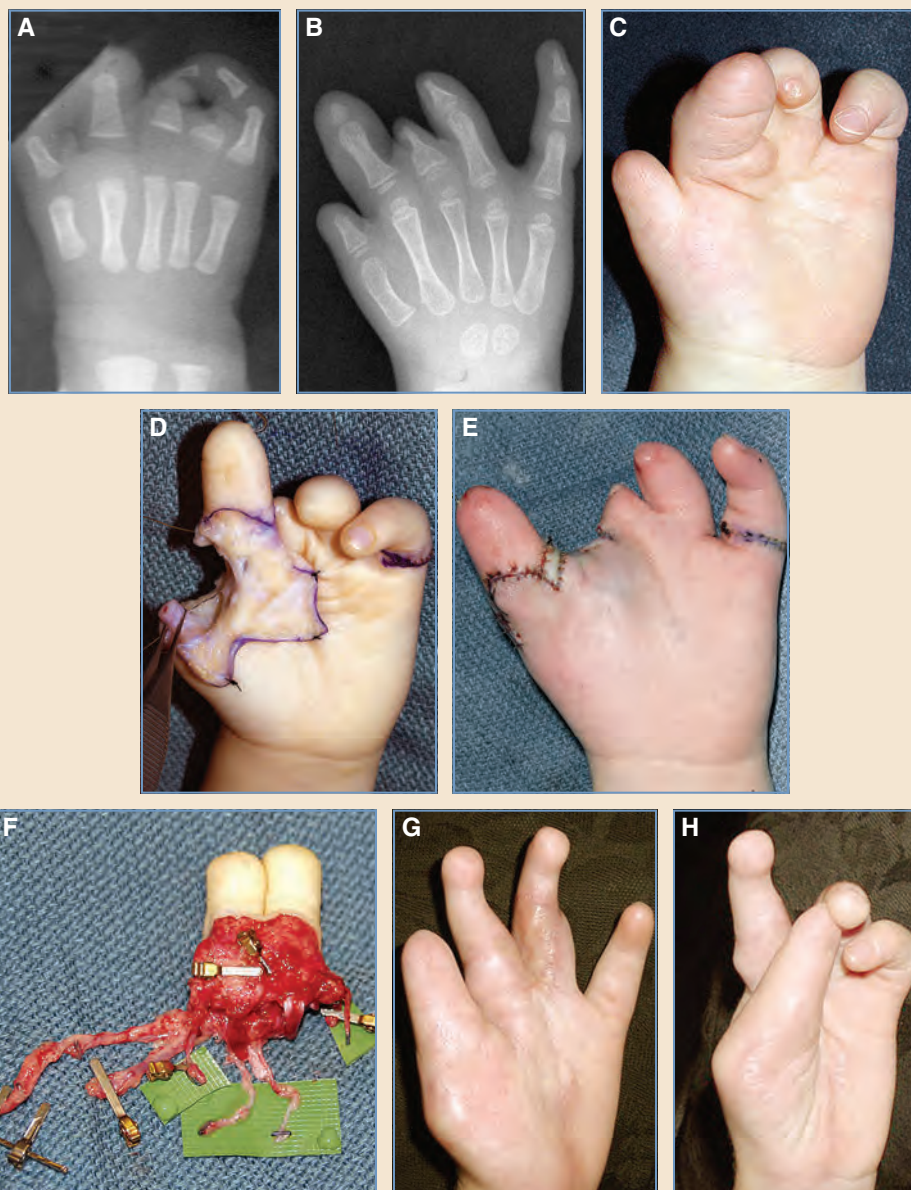


Fig. 43-50 Thumb in constriction ring syndrome. **A**, This radiograph of a child with acrosyndactyly following release shows congenital amputations of the thumb at the IP joint level, the index and ring digits through the middle phalanx, and the long digit through the proximal phalanx. **B** and **C**, A radiograph and clinical appearance of the same hand 18 months later. **D** and **E**, The index phalanges were transferred on their vascular pedicle as a pollicization to lengthen the thumb, which already had a good complement of thenar intrinsic muscles. The constriction ring of the fifth digit was corrected. Skin grafts were needed on the ulnar side of the new thumb. **F**, When the patient was 3 years of age, a double second and third toe composite transfer was performed on top of the long and ring digits. A syndactyly release was performed 3 months later. **G** and **H**, At 6 years of age, this girl is a very confident first grader with a mobile thumb and a strong pinch.

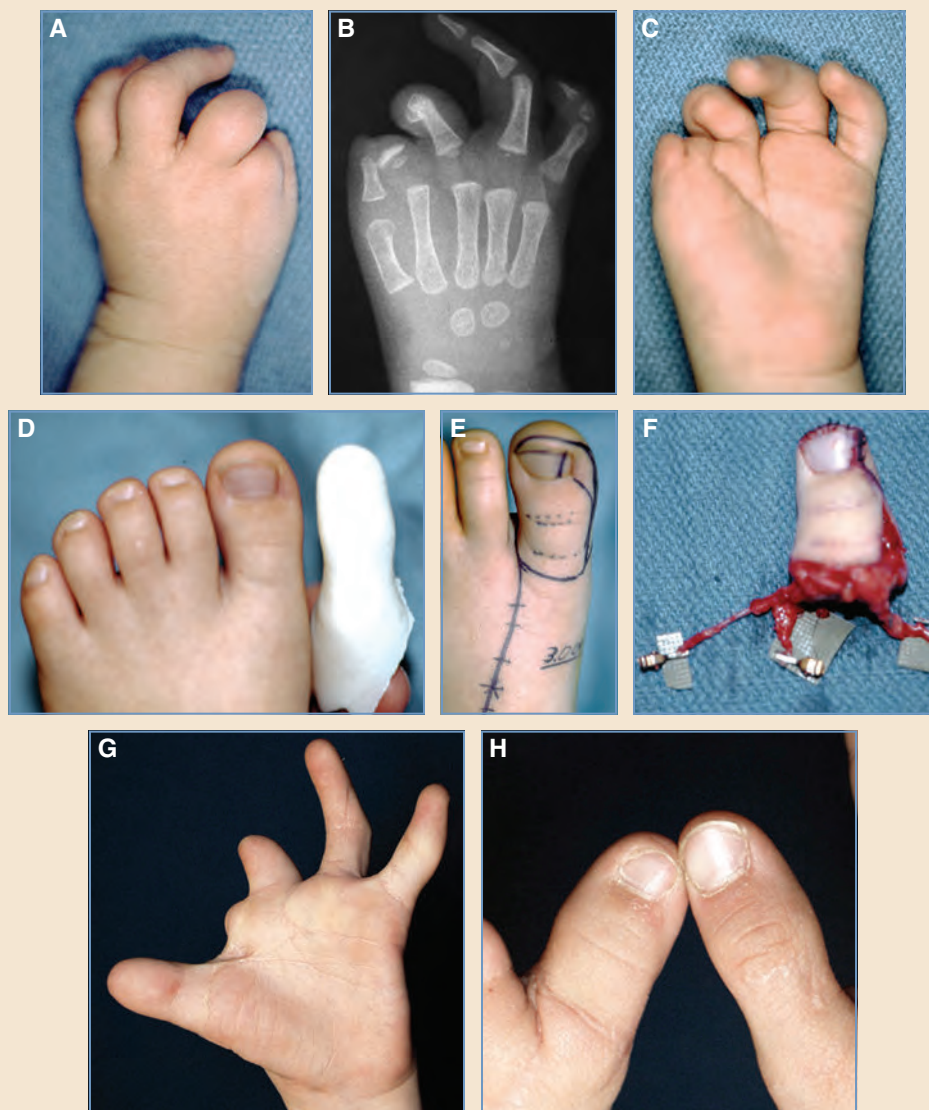


Fig. 43-51 Thumb with constriction ring syndrome treated with microvascular toe transfer. **A-C**, This boy, born with acrosyndactyly, had thumb loss at the interphalangeal joint level, no first web space, and congenital amputations of the index and long digits at the phalangeal level. The connecting bands present at birth were released in the newborn nursery. His first surgery was a distally based radial forearm fasciocutaneous flap to the first web space and release of the camptodactyly of the ring finger. **D-F**, Thumb length was augmented with a modified great toe transfer. A methylmethacrylate mold of the opposite normal thumb was used to measure and contour the larger great toe at the time of operation. The incision markings are shown on the ipsilateral great toe, and the specimen is seen in transit to the recipient left thumb. **G** and **H**, Four years later. Thumb sensation is normal, and motion measures 30 degrees at the IP joint and 50 degrees at the MP joint. The contour has been excellent. No revisions have been performed.

In several children we have first lengthened a deficient metacarpal with an intact CMC joint to provide a good foundation for second toe transfer. The keys to this procedure are to proceed slowly, to minimize soft tissue dissection, and to avoid injury to growth centers. The major problem with any toe transfer is the availability of toes, which, like the fingers, may be hypoplastic or aplastic.

Various types of on-top-plasties have been described in the hand literature; these consist of transfer and distal advancement of composite soft tissue and bone segments from adjacent fingers. We prefer free transfers instead of these local transfers, which tend to require extensive dissection and lead to secondary contractures within the first web space (see Fig. 43-50).

TYPE IX THUMB HYPOPLASIA: FIVE-FINGERED HAND

Anatomy

In type IX hypoplasia the thumb is smaller in width and longer in length and has the characteristics of a finger. As the radial border digit, it lies in the same plane as the ulnar four digits and is nonopposable. It is usually the same length as the adjacent index finger. The digit is slender and may often be joined to the index finger in an incomplete simple syndactyly. A severe deficiency or nonexistent first web is often present. The skeletal anatomy is similar to that of the index ray. A metacarpal with a distal growth center and three phalanges with proximal growth centers are present. The scaphoid is usually absent or hypoplastic.¹²⁵ The thenar musculature (AbPB, FPB, OP muscles) is absent, as is the adductor pollicis (AddP). Instead, the usual digital intrinsic muscles are present: a lumbrical, palmar interosseous, and dorsal interosseous muscle (Fig. 43-52). The extrinsic flexors and extensors mimic those of the normal fingers. Because the radial digit lies in the same plane as the other fingers in the hand, manipulation of objects is usually performed by using lateral scissoring between the first two digits and between the second and third if a first web syndactyly is present. If first web syndactyly is left untreated, patients tend to attenuate the transverse metacarpal ligament and to “autopollicize” into an abducted and slightly pronated posture.

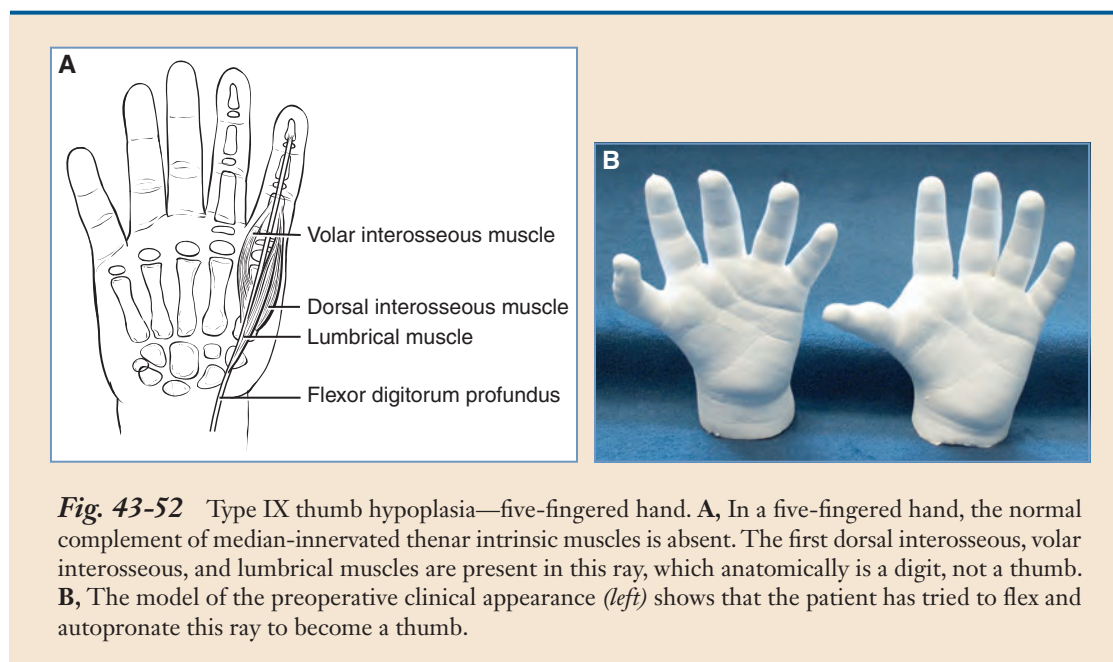


Fig. 43-52 Type IX thumb hypoplasia—five-fingered hand. **A**, In a five-fingered hand, the normal complement of median-innervated thenar intrinsic muscles is absent. The first dorsal interosseous, volar interosseous, and lumbrical muscles are present in this ray, which anatomically is a digit, not a thumb. **B**, The model of the preoperative clinical appearance (*left*) shows that the patient has tried to flex and autopronate this ray to become a thumb.

Management

The optimal management of these patients is to pollicize the radial digit. The technique employed is similar to that for thumb aplasia (type V).^{83,90,93} Like pollicization for thumb aplasia, rebalancing of the intrinsic musculature is paramount to the success of the procedure. In the five-finger hand anomaly, the dorsal interosseous to the radial digit is only unipennate, and secondary opponensplasty with an abductor digiti quinti muscle transfer is occasionally necessary.



Fig. 43-53 The thumb in a five-fingered hand. **A** and **B**, The right hand of a child with bilateral five-fingered hands; she has autorotated the radialmost digit into a pseudothumb position in an effort to function more effectively. **C** and **D**, In one procedure, the thumb was shortened and fused at the proximal interphalangeal joint, and a first web space was created with a radial forearm fasciocutaneous flap, which is seen before, during, and 5 years after the procedure. A superficial flexor tendon was transferred to improve palmar abduction (opposition). **E** and **F**, Five years postoperatively, there has been no contracture or diminution of this web space, which was lined with full-thickness flap tissue.

When type IX hypoplasia is seen late, two other options are available: construction for a first web space with a distally based forearm flap or free tissue transfer plus arthrodesis of the PIP joint, or a rotation-recession osteotomy of the first metacarpal with pollicization and intrinsic rebalancing (Fig. 43-53). This condition should not be confused with a triphalangeal thumb, in which the thumb is somewhat shorter than the other digits. Although an extra phalanx is present by definition, extrinsic and intrinsic muscle anatomy mimics a thumb more than a finger. The extra phalanx is commonly angulated. This is commonly associated with radial polydactyly.

TYPE X THUMB HYPOPLASIA: RADIAL POLYDACTYLY

Anatomy

Despite the level of duplication, each partner thumb is hypoplastic to varying degrees, with the radial duplicate usually being the most severely affected. The complexity of the deformity and therefore the surgical correction required increases as the level of the duplication progresses proximally. Associated triphalangia of the ulnar partner further complicates the surgical reconstruction. Specific abnormalities in each duplicate are seen in the nail plate, the osteoarticular column, and both the intrinsic and extrinsic myotendinous units of each thumb. The nail plate of each duplicate is always more narrow than that of the unaffected thumb. The entire skeletal ray is hypoplastic in duplications proximal to the MP joint. Myotendinous abnormalities are common, with the extrinsic extensor being almost universally shared.

Deviation of the partners toward each other indicates abnormal insertions of the extrinsic tendons into the distal phalanges. Connections between the extrinsic flexors and extensors are not unusual. These tendinous interconnections will limit function and cause digital angulations. The first web is usually unaffected in duplications involving the distal phalanx, but as the level of duplication lies more proximal, the first web space becomes increasingly deficient.

Management

The management of patients with duplications of the thumb will vary for the level of duplication and are discussed in more detail elsewhere in this book (see Chapter 46). In general, the surgeon should use the best parts of each thumb to construct the best possible thumb. Distal duplications (types I and II) are usually managed by preservation of the ulnar duplicate with extra soft tissue being provided from the radial thumb. The resulting nail is smaller than that on the normal side. The Billhaut-Cloquet procedure is avoided if possible because of the predictable problems of nail ridging and diminished IP joint motion.⁸² Duplications at the proximal phalangeal level (types III and IV) are also commonly managed with ablation of the radial partner and closure as required with tissue from this thumb. Myotendinous abnormalities need to be addressed with the division of interconnections and centralization of insertions. Advancement of the detached thenar intrinsics into the retained extensor mechanism is mandatory. Collateral ligament preservation and reattachment is key in providing a stable MP joint.¹²⁶ The metacarpal head will be broad in these patients and require trimming on the redundant radial side. A closing wedge osteotomy of the metacarpal may also be required to realign the newly constructed thumb. Appropriate trimming of excess skin should result in wound closure, which is positioned in the high midaxial position. Duplication in the metacarpal shaft (type V) can often be managed by simple ablation

of the radial partner and concomitant manta ray flap⁵⁹ or four-flap Z-plasty for any first web deficiency. Duplications at the CMC joint level (type VI) and triphalangeal varieties all need to be dealt with on their merit. Digital transposition of the ulnar thumb onto the retained base of the radial partner is not unusual. Skeletal realignment, tendon repositioning, and first web releases can usually be completed in a single stage.

TYPE XI THUMB HYPOPLASIA: SHORT SKELETAL RAY

Anatomy

Deficiencies of the osteoarticular column of the thumb may result in a short, hypoplastic thumb.⁶⁶ Bony abnormalities can occur as isolated findings (brachymetacarpia, brachyphalangia), all bones in combination, or as part of a generalized syndrome, such as the acrocephalosyndactyly syndromes (for example, Apert, Pfeiffer, Carpenter) or the Rubinstein-Taybi syndrome. Joint function is usually impaired on either side of the abnormal bone (or bones). In patients with anomalies of a single bone, brachymetacarpia or brachyphalangia, the remaining components of the thumb tend to be unaffected, and in patients with generalized syndromes, other abnormalities of the thumb components are common. In the acrocephalosyndactyly (ACS) syndromes, delta phalanges are usual. This typically involves the proximal phalanx, with a longitudinal epiphyseal bracket on the radial side. This abnormal growth plate checkreins growth on the radial side and results in a radial clinodactyly of the thumb. The metacarpal is usually short, and the distal phalanx is short and broad. Many surgeons think that the abnormal proximal phalanx and broad distal phalanx are variations of duplication. Myotendinous anomalies are associated with poor joint function. Deficiencies of the first web are most commonly seen in patients with Apert syndrome. This can be anywhere along a spectrum of mild adduction contracture to complex syndactyly involving the first two rays (Fig. 43-54).

The whole spectrum of patients with metabolic bone diseases, skeletal dysplasias, and tumors may include thumb hypoplasia. In general, very little surgical correction is required for these children.

Management

Many patients with short thumbs that are not associated with generalized conditions do not require any treatment, since the length deficiency is mild. In thumbs that do have an obvious length deficiency, distraction lengthening with a secondary bone graft to the subsequent bony defect at the metacarpal level is the treatment of choice.

Patients with generalized syndromes require a multidisciplinary approach to the management of that thumb, and hand procedures can be coordinated with other required treatments. Children with complex hand anomalies, such as those with Apert syndrome, require special attention to optimize the function in their hands. First web syndactyly needs to be managed in the first 3 to 6 months of life so that these children can develop with their thumbs released before the development of prehension. The deficiency of the first web in the majority of the children with cra-

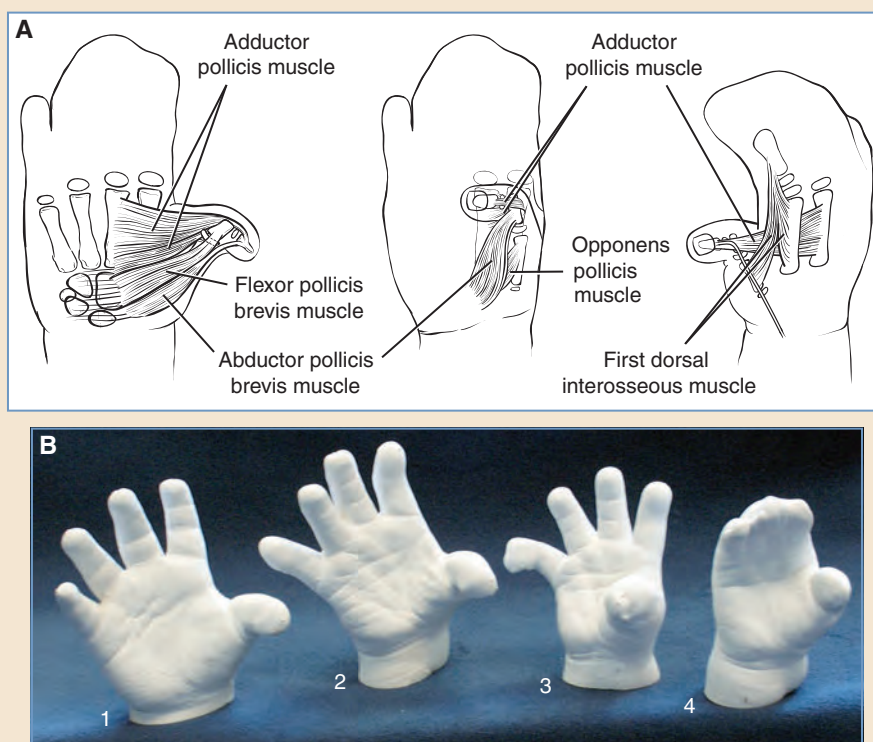


Fig. 43-54 Type XI thumb hypoplasia—short skeletal ray. **A**, A short thumb with or without radial clinodactyly is seen in many syndromes. The primary osseous abnormalities are usually seen at the proximal phalangeal joint level. The median- and ulnar-innervated intrinsic muscles are all present and are very hypertrophied. Extrinsic and intrinsic flexors and extensors are normal up to the phalangeal level. **B**, The thumbs of children with four different syndromes all look similar: 1, Rubinstein-Taybi; 2, Pfeiffer; 3, Greig cephalopolysyndactyly; and 4, Apert acrocephalosyndactyly.

niosynostoses can be adequately treated with a manta ray flap or a four-flap Z-plasty. In the more complex type II and III hand anomalies in Apert syndrome, where there is complete syndactyly of the first web, we have found tissue expansion of the first web to be a very satisfactory means of producing a long-lasting release of the first web.³³ The deficiencies of length and angulation in these complex anomalies that are associated with a “delta” phalanx should be managed with an opening wedge osteotomy and iliac crest bone graft, or in Apert syndrome, the synostotic bone from the base of the fourth and fifth metacarpals. Because these bones are quite small, this procedure is usually delayed until 4 or 5 years of age^{66,82} (Fig. 43-55).

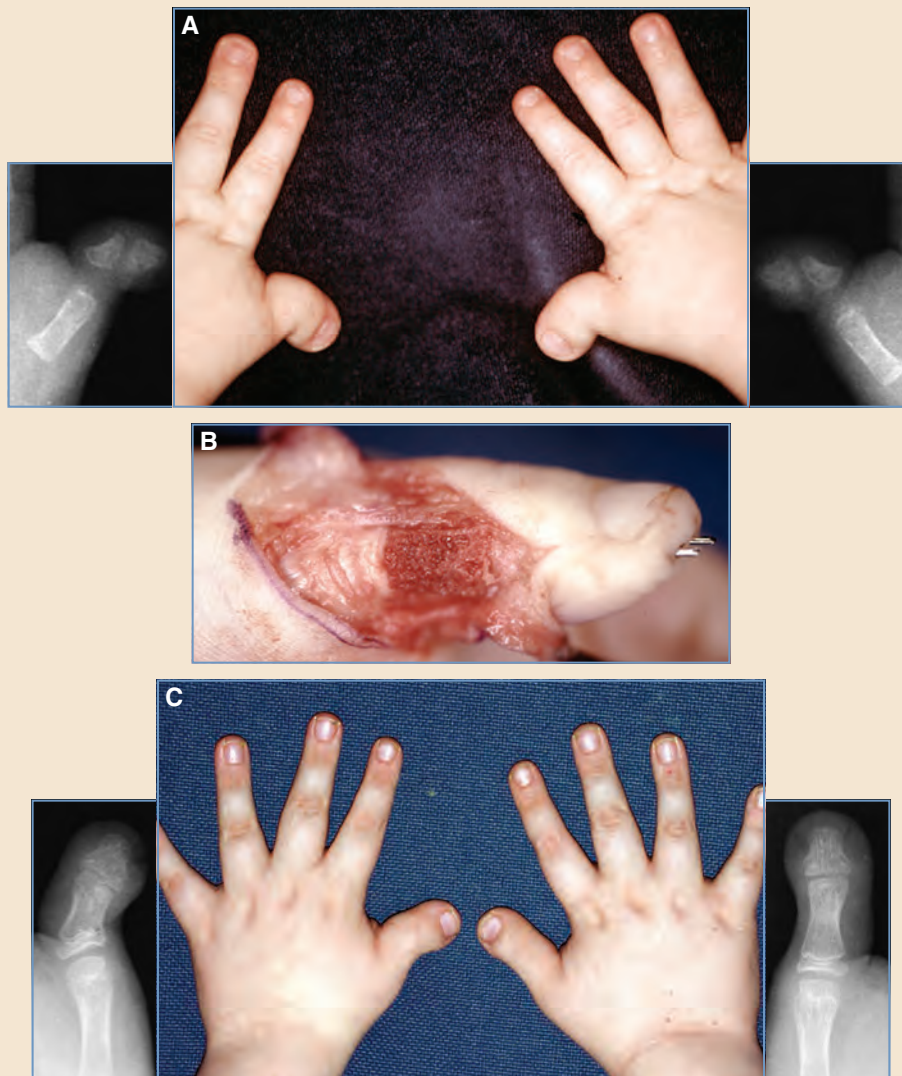


Fig. 43-55 Short thumb in syndromic patients. **A**, Preoperative appearance and thumb radiographs of a young boy with bilateral radial clinodactyly of the thumbs (“hitchhiker thumbs”) associated with Rubenstein-Taybi syndrome. **B**, The soft tissue was lengthened on the radial side with a large Z-plasty and a composite graft. The bone was lengthened, as seen here, with an opening wedge osteotomy and corticocancellous bone graft held with two C-wires. **C**, The same thumbs are seen 7 years later. The right side has grown normally, and on the left, the deformity has recurred because of the premature closure of the radialmost border of the growth plate.

KEY POINTS

- Congenital thumb differences are very common and are present to some degree in most syndromic categories, including constriction ring syndrome, symbrachydactyly, typical cleft hand, and radial polydactyly, among others.
- There is a wide spectrum of anatomic and functional deficiencies.
- A careful physical examination and identification of all soft tissue and skeletal deficiencies is the most important component in planning management of these children.
- The original German classification, which outlines five categories (types I through V), is most commonly used in clinical management.
- A moderate-to-severe deficiency of the first web space is the most commonly missed deficiency in these children. A simple release with local flaps is one of the most functional procedures done in pediatric hand surgery.
- Despite impressive refinements in microsurgery, pollicization is still the procedure of choice for the type IIIB thumb.

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44

Congenital Hand Anomalies: Failure of Differentiation

Robert J. Havlik • Patrick C. Hettinger



The American Society for Surgery of the Hand and the International Federation of Societies for Surgery of the Hand have developed a classification system for congenital anomalies of the upper extremity. In this system, failure of differentiation includes a broad group of disorders, including those involving soft tissue, those involving skeletal tissue, and congenital tumorous conditions. Failure of differentiation refers to anomalies in which the basic units have developed, but the final form is not completed appropriately (Boxes 44-1 and 44-2).

Within this broader group of disorders, syndactyly is the diagnosis of most interest to plastic surgeons. Most of this chapter focuses on the characteristics and the treatment of this anomaly. The other diagnoses considered failures of differentiation occur less frequently and the discussion of these is more limited in this chapter.

Box 44-1 Classification of Congenital Hand Anomalies: Failure of Differentiation (Separation) of Parts

- | | |
|---|---|
| <p>A. Soft Tissue Involvement</p> <ol style="list-style-type: none"> 1. Disseminated <ol style="list-style-type: none"> a. Arthrogryposis (including multiplex congenita) <ol style="list-style-type: none"> (1) Severe (2) Moderate (3) Mild 2. Shoulder <ol style="list-style-type: none"> a. Undescended Shoulder <ol style="list-style-type: none"> (1) Sprengel shoulder b. Absence of thorax muscles (including Poland syndrome) <ol style="list-style-type: none"> (1) Pectoralis major (2) Pectoralis major and minor (3) Other 3. Elbow and Forearm <ol style="list-style-type: none"> a. Aberrant muscle <ol style="list-style-type: none"> (1) Aberrant muscles of the long extrinsic flexors (2) Aberrant muscles of the long extrinsic extensors (3) Aberrant intrinsic of the hand (4) Other 4. Wrist and Hand <ol style="list-style-type: none"> a. Cutaneous syndactyly (complete and incomplete) <ol style="list-style-type: none"> (1) Radial (first interdigital space) (2) Central (second/third interdigital space) (3) Ulnar (third interdigital space) (4) Combination of (1) ± (2) or (3) b. Congenital flexion contracture (camptodactyly) <ol style="list-style-type: none"> (1) Fifth digit (2) Other c. Thumb-in-palm deformity d. Deviated finger without bony deformity (laxity secondary to differentiation of muscle or capsule) <ol style="list-style-type: none"> (1) Radial or ulnar <ol style="list-style-type: none"> (a) Isolated digit (b) Congenital ulnar drift (windblown hand) 5. Skin and Appendages <ol style="list-style-type: none"> a. Pterygium (webbing) of the axilla or elbow b. Cutis aplasia congenita c. Congenital clubbing of the nails d. Tusk nail deformity, palmar nail e. Other | <p>B. Skeletal Involvement</p> <ol style="list-style-type: none"> 1. Shoulder <ol style="list-style-type: none"> a. Congenital humeral varus b. Other 2. Elbow <ol style="list-style-type: none"> a. Elbow synostosis <ol style="list-style-type: none"> (1) Humeroradial (2) Humeroulnar (3) Total elbow b. Elbow ankylosis (joint segmentation present) 3. Forearm <ol style="list-style-type: none"> a. Proximal radioulnar synostosis <ol style="list-style-type: none"> (1) Without radial head dislocation (2) With radial head dislocation b. Distal radioulnar synostosis 4. Wrist and Hand <ol style="list-style-type: none"> a. Synostosis of carpal bones <ol style="list-style-type: none"> (1) Lunotriquetral synostosis (2) Capitohamate synostosis (3) Scapholunate synostosis (4) Others b. Synostosis of the metacarpal bones <ol style="list-style-type: none"> (1) Ring-small synostosis (2) Others c. Synostosis of the phalanges (osseous syndactyly, complex syndactyly) <ol style="list-style-type: none"> (1) Radial (first-second rays) (2) Central (second-third, third-fourth rays) (3) Ulnar (fourth-fifth rays) (4) Mitten hand (including Apert hand) (5) Other d. Symphalangia <ol style="list-style-type: none"> (1) Proximal interphalangeal (PIP) joint (2) Other e. Congenital deviation (clinodactyly) <ol style="list-style-type: none"> (1) Idiopathic clinodactyly <ol style="list-style-type: none"> (a) Fifth finger (b) Thumb (including a delta phalanx) (c) Others f. Hypersegmentation <ol style="list-style-type: none"> (1) Triphalangeal thumb (2) Others |
|---|---|

C. Congenital Tumorous Conditions

1. Vascular system
 - a. Hemangioma
 - b. Malformations
 - (1) Capillary
 - (a) Portwine stain
 - (b) Others
 - (2) Venous
 - (3) Venolymphatic
 - (4) Arterial (including arteriovenous fistulas)
 - (5) Lymphatic
 - (6) Others
2. Neurologic
 - a. Neurofibromatosis
 - b. Neuroblastoma
 - c. Others
3. Connective Tissue
 - a. Juvenile aponeurotic fibroma
 - b. Other
4. Skeletal (not including overgrowth syndromes)
 - a. Osteochondromatosis (including multiple hereditary exostosis)
 - b. Enchondromatosis
 - c. Fibrous dysplasia
 - d. Epiphyseal abnormalities
 - e. Other

Box 44-2 Failure of Differentiation Disorders**Soft Tissue Disorders**

Arthrogryposis
 Thumb-in-palm deformity
 Congenital trigger digit or thumb
 Syndactyly
 Poland syndrome
 Camptodactyly
 Aberrant muscles of the elbow and forearm
 Sprengel deformity

Skeletal Disorders

Elbow synostosis
 Radioulnar synostosis
 Synostosis of the carpal bones, metacarpal bones, and phalanges
 Symphalangism
 Clinodactyly
 Triphalangeal thumb

Congenital Tumorous Conditions

Hemangiomas
 Venous malformations
 Lymphatic malformations
 Arterial malformations
 Neurofibromatosis
 Neuroblastoma
 Osteochondromatosis
 Enchondromatosis and fibrous dysplasia

Failure of Differentiation: Soft Tissue

Arthrogryposis

Arthrogryposis is a diverse group of disorders sharing common clinical characteristics. They are discussed in Chapter 50 and will not be discussed here.

Congenital Flexion Deformities of the Hand

A congenitally flexed position of the thumb may be a normal variant in an infant, but it may also be seen in many disorders, including congenital trigger thumb, congenital clasped thumb, and arthrogryposis. A physical examination is critical for establishing a proper diagnosis and guiding treatment, with detailed attention to the hand and the associated findings and syndromes. On the hand, the presence or absence of flexion and extension creases should be determined. These findings can indicate that a digit has never been flexed or extended. They also help to identify the level at which the flexion deformity of the thumb occurs. In many children, the long extensors of the thumb undergo a delay in maturation, leading to extensor weakness and resultant limited or absent thumb extension and abduction during early infancy, with flexion deformity at the level of the metacarpophalangeal (MP) joint. Children with congenital trigger thumb have a flexion deformity at the interphalangeal (IP) joint of the thumb. In contrast, children with congenital clasped thumb deformity have flexion at the MP joint level.

In children with apparently normal development but without thumb extension and abduction, the only treatment usually necessary is splinting the thumb in both extension and abduction. A forearm-based splint should be fabricated as soon as a deficiency in motion is noted. It should be worn 23 hours a day, allowing time for bathing and hygiene. Most of these patients respond favorably and completely as maturation occurs.

Congenital trigger thumb is common and relatively easily differentiated from congenital clasped thumb based on the level of the deformity. A trigger thumb is typically held in a flexed position at the joint, unlike the MP joint in a normal hand and the congenital clasped thumb noted later in the chapter. A trigger thumb may be truly congenital and evident at birth, or it can arise during early infancy. Once the digit is flexed, the child will generally hold it in this position, because the sensation of the triggering may be uncomfortable. Less frequently, the digit is held in extension at the IP joint of the thumb. A nodule (Notta node) may be palpable just proximal to the thumb flexion crease. The role of splinting in this disorder is controversial; some authors recommend it, and others think it is not beneficial. Treatment is guided by the knowledge that, in some series, the disorder spontaneously resolved.^{1,2} Surgery should be deferred until the child is at least 9 to 12 months of age to allow spontaneous resolution. If the lesion persists, surgery is carefully performed to prevent injury to the digital sensory nerves that lie more centrally in the thumb relative to the other digits. A transverse incision is made at the proximal thumb palmar flexion crease just through the dermis. The digital sensory nerves are identified, and the A1 pulley is completely released along the radial aspect. Full excursion of the thumb without restriction should be demonstrated before closure.

The treatment for flexion deformity at the MP joint depends on associated findings. As noted previously, if the child is otherwise normal, the flexion deformity may result from a simple delay in maturation and extensor system weakness, which will respond to splinting. These children need continued follow-up, because the flexed posture may indicate a congenital clasped thumb.

Congenital clasped thumb can be categorized into two groups: supple and complex.³ The supple subtype often has only a deficiency of the extensor mechanism, whereas the complex type also has a variable degree of MP joint contracture, collateral ligament abnormality, thenar muscle hypoplasia, web space tightness with skin deficiency, and flexor pollicis longus tightness. Complex congenital clasped thumb can be an isolated deformity, but it often occurs in association with other deformities, including arthrogryposis. Clasped thumb deformity associated with arthrogryposis is discussed in Chapter 50.

Syndactyly

Syndactyly is derived from the Greek “syn,” meaning *together*; and “dactylos,” meaning *digits*. Syndactyly is one of the most common congenital anomalies involving the hand and upper extremity.⁴ In most large series of congenital anomalies of the upper extremity, the three most common anomalies of the upper extremity are syndactyly, polydactyly, and camptodactyly.⁵ The differences in the incidence of these three anomalies may largely reflect the patient series and the population studied, rather than a true variability in the overall incidence.⁵ For example, polydactyly is much more common among African-Americans, in whom it is transmitted in autosomal dominant fashion. Overall, the incidence of syndactyly in the general population is estimated to be one in 2000 births.^{4,6,7} In England and Wales,⁸ where the British National Health Service provides comprehensive population-based health statistics, the incidence of syndactyly in the British population is approximately one in 1725 births. In more than 95% of all pregnancies carried to term in the United States, the infant is “normal,” and the most common major congenital anomaly is orofacial clefting (cleft lip and/or cleft palate), which has an incidence of approximately one in 700 births.⁹ If the incidences of syndactyly, polydactyly, and camptodactyly are nearly equal, then their combined incidence is roughly the same as that of orofacial clefting.

Many classification schemes have been proposed for syndactyly. Some group entities together and some further divide them into subcategories. One of the most widely used is a simple four-category system. In this classification scheme, syndactyly is characterized as *complete* or *incomplete*, based on the extent of webbing present between the digits, and as *simple* or *complex*, based on the presence or absence of bony union. In complete syndactyly, the webbing extends all the way distally to involve the fingertips, whereas in incomplete syndactyly the webbing extends to any-



Fig. 44-1 A, Simple complete syndactyly. B, Simple incomplete syndactyly. C, Apert rosebud type III hand with complex syndactyly (see Apert Syndrome: Acrosyndactyly section later in the chapter).

where between the fingertips and the normal position of the web space (Fig. 44-1). Although the normal position of a web space varies, generally, the most distal aspect of the web space lies on the palmar aspect of the hand, at a point halfway between the metacarpal head and the proximal phalangeal head.⁷ *Simple syndactyly* refers to cases in which the digits are joined by only the skin and fibrous tissue of the hand, whereas *complex syndactyly* also involves a coalition between the bones of the phalanges (see Fig. 44-1). The usefulness of this classification system lies both in its simplicity and its large descriptive groups, which are helpful to many physicians.

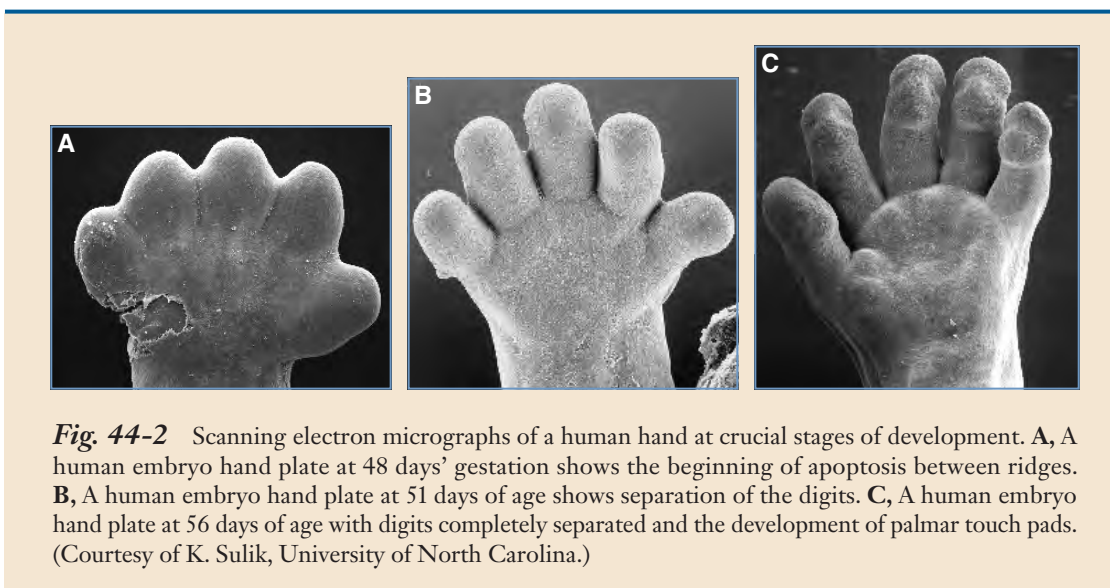
As a research classification system or as a system of precise communication between physicians who treat these disorders frequently, this rudimentary classification system is less useful. Syndactyly can vary tremendously across a spectrum, sometimes involving abnormal connections between tendons, nerves, and arteries. Surgeons need to understand that these abnormal connections occur, recognize them, and define and treat them accordingly. Syndactyly can also be a manifestation of other syndromes, such as Apert syndrome or Poland syndrome. In pseudosyndactyly, the digits were once completely separated but are drawn back together, as in constriction ring syndrome (amniotic band syndrome) or dystrophic epidermolysis bullosa. These special circumstances will be discussed separately later in the chapter. The following discussion pertains to nonsyndromic syndactyly.

SYNDACTYLY

Genetics, Etiologic Factors, and Embryology

Syndactyly occurs in approximately twice as many males as females and most often involves the long-ring web space of the hand and the second-third web space of the foot. The long-ring web space is affected in 50% of all cases.⁷ The ring-small web space is involved in 30% of cases, the index-long web space is affected in 15% of cases, and, generally, the thumb-index web space is spared, involved in only 5% of cases.⁴ Approximately half of all cases of syndactyly are bilateral. Most are sporadic, isolated anomalies. However, 10% to 40% are inherited on a familial basis. The large variation in familial inheritance is probably dependent on the population sample studied. Flatt⁴ reported a familial incidence of 40% in his population of 413 patients with syndactyly. This relatively high rate of familial occurrence may be related to the large, stable population in Iowa that made up his reference population. All patterns of genetic inheritance have been postulated based on retrospective pedigree analysis, and the most common pattern is autosomal dominant with incomplete penetrance.⁶

In normal development, the hand plate develops from approximately week 5 to week 7 of gestation.^{5,10,11} Hand development is driven by diffusion of growth factors in local tissues, creating local concentration gradients. The hand plate develops five ridges that are the digital precursors. These ridges develop by programmed cell death, or apoptosis, of the apical ectodermal ridge in the areas between the ridges, typically at 48 days of gestation^{5,11} (Fig. 44-2). The process of apoptosis starts distally and progresses proximally. Concurrently, the mesenchymal tissue in the ridges and digital buds undergoes continued condensation and growth. Digital separation is completed by programmed cell death of the intervening tissue, typically at 51 days of gestation¹¹ (see Fig. 44-2). By 8 weeks of age, the digits have developed touch pads distally, and the hand's appearance is largely defined¹¹ (see Fig. 44-2). The failure of separation of the digital rays causes syndactyly. This pattern of morphogenesis may account for the relatively low incidence of involvement of the first web space, because the thumb ray is known to separate from the rest of the hand earlier in this process of differentiation. However, we do not know how this mechanism leads to the male



predominance of the disorder or the predominant site of involvement—the first postaxial web space in the foot and hand (the second-third toe space and the ring-long web space of the hand).

Functional Implications and Timing of Surgery

Depending on its location within the hand, syndactyly may have limited impact on overall hand function and development. Therefore the timing of separation of the digits in syndactyly depends on the digits that are affected and the extent of overall involvement of the hands. Developmentally, the ability to position the hand in space (a cortical function) and the development of grasp are established by a year of age.¹² The ability to position the hand and use effective grasp undergoes continued refinement to 3 years of age. Therefore, in cases involving the thumb and index finger, separation of the digits and the creation of a sufficient first web space are desirable at an early age (younger than 6 months). Cases that involve the ring-little or thumb-index web spaces (border digits) require earlier separation because of the inherent length discrepancy between the thumb and index fingers and the ring and little fingers. Failure to separate these digits before 6 months of age can lead to permanent distortion of the longer finger (index finger and ring finger, respectively) as it is held back by the tethering force of the shorter finger. In complex syndactyly, in which the digits are conjoined with bony fusion between the distal phalanges, earlier separation is needed to prevent deformation of the digits with growth. For hands with multiple digital involvement, planning an appropriate course of treatment is imperative. Treatment should allow timely separation between border digits and completion of the process within the first few years of life. This is especially true in Apert hands, in which digital separation must be coordinated with craniofacial operations. An additional advantage of early surgery in these complex cases is that bilateral surgery can be performed and is usually well tolerated until patients are 1 year of age. This minimizes the number of anesthetics and independent operative procedures that a child must undergo.

In contrast, separation of the most common digital syndactyly involving the long finger and ring finger web space can be deferred until the child is older without significant functional sequelae. However, social issues begin to become a significant issue by school age. Separation should be completed before this to prevent ridicule and social ostracism.

Goals and Principles of Surgery

Three primary goals are considered when planning the approach for reconstructing a hand with syndactyly: (1) allow the patient to position the hand (and fingers) in space; (2) provide adequate and sensate skin coverage; and (3) provide a satisfactory power grasp with the ability to handle objects precisely.⁴ Six primary tenets of reconstruction of syndactyly help to ensure that these goals are met:

1. All osseous and soft tissue between the digits should be separated.
2. The nerves and the vascular supply are optimally preserved.
3. An adequate web space is established between the digits by developing a flap of appropriate length that is inset without tension.
4. Interdigitating flap coverage is provided over the mid-lateral digit, ideally at the level of the PIP joint.
5. Appropriate distal phalanges are developed.
6. Perforated, full-thickness skin grafts are used to minimize the effects of secondary wound contraction on the lateral aspect of the phalanges.

The treatment paradigm for the separation of complete and incomplete syndactyly is very similar in all digits, except for the thumb-index web space, which is discussed separately.

Separation of the digits includes separation of the conjoined dorsal and palmar skin, subcutaneous fat, conjoined ligaments, and sites of osseous union, while preserving the integrity of the nerves and blood vessels. When separating the skin, care is taken to optimize the digital coverage

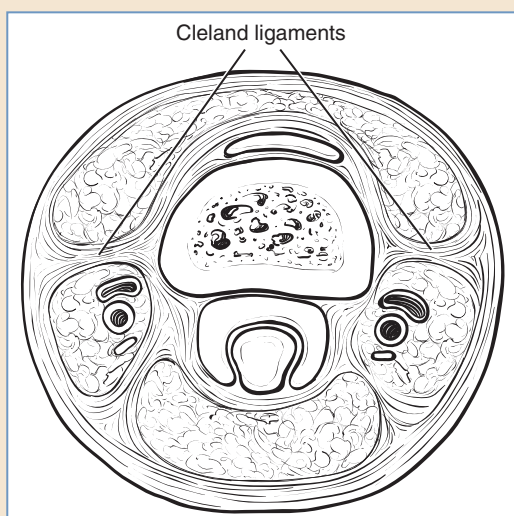


Fig. 44-3 Cross-section of a digit showing the location of Cleland ligaments.

with skin and to resurface the web space, as described later in the discussion. The subcutaneous fat is usually moved en bloc with the overlying skin. In virtually all syndactyly, simple or complex, complete or incomplete, conjoined ligaments exist between the digits. These are probably conjoined sections of Cleland ligaments (Fig. 44-3). In normal digits, Cleland ligaments usually provide points of fixation of the skin to the underlying skeleton of the digit in the mid-axial line of the digit. This provides stability of the skin during grasping and digital manipulation, and it transfers the force imparted by the musculoskeletal units to grasp the object in hand and resist shear stresses. In syndactyly, the coalition between these structures results in the strong fibrous union between the skeletal framework of the digits, and this must be divided. Complete separation of these ligaments is essential, with preservation of the underlying neurovascular bundles. It is seldom, if ever, necessary to extend the division of these deep soft tissues more proximally to include the division of the transverse metacarpal ligament.

In normal anatomic relationships, the neurovascular bundles lie just palmar to this area of ligamentous union. Separation of the neurovascular bundles requires meticulous attention to the anatomy. Distal bifurcations of the blood vessels may be encountered, and if they are too distal to allow appropriate separation of the digits, two precautions are appropriate:

1. It is generally accepted that only one side of a digit be separated at each operative session. Surgeons should not attempt to separate both sides of a digit for concern of vascular disruption or vascular spasm.
2. If the vascular integrity of a digit is in question, a microvascular clamp should be placed distal to the bifurcation of the digital artery at the contemplated site of division of the digital artery and the tourniquet released. Vascular perfusion to the digit should be sufficient before the proper digital artery is ligated.

In contrast, nerves generally pose less of a problem than the vascular supply. A distal branching of the digital nerves can be separated bluntly with the back of a scalpel blade or the closed points of tenotomy scissors. Appropriate length of the nerves can almost always be gained by these relatively simple techniques.

Creating an appropriate web space and commissure between digits is essential for separating digital syndactyly. Flaps are always preferable to grafts in this location for their suppleness and pliability. The web space must be soft and supple to facilitate digital motion without irritation from unstable skin coverage. In addition, placing a soft, supple flap of tissue will minimize the chance of distal migration of the web space with further growth of the hand. Several flap designs for creating and resurfacing the web space have been promoted over the past 100 years, includ-



Fig. 44-4 Dorsal hand flap design extending two thirds of the distance between the metacarpal heads and the PIP joint.

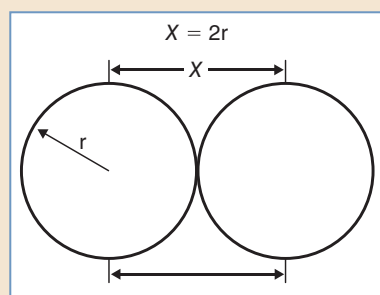


Fig. 44-5 Separation of syndactyly always involves a net skin deficiency. The available skin measures only four times the radius ($2r$ above and $2r$ below), whereas the surface area to be covered equals $2\pi r$ (πr on the medial aspect of each finger).

ing dorsal flaps, palmar-based flaps, and various combinations of these flaps in interdigitating patterns. Ideally, the web space has a dumbbell-shaped surface, with a narrower waist and wider dorsal and palmar components. It should be sloped from proximodorsally to distally in the palm, replicating the normal anatomy. Perhaps the most widely used design is that of a dorsal web flap described more than 50 years ago by the Indiana plastic surgeons Bauer, Tondra, and Trusler¹³ (Fig. 44-4). This technique has the advantages of creating a flap of appropriate width, length, and dorsopalmar slope in a reliable manner and of leaving the web space largely devoid of scar tissue that could create secondary contracture. The disadvantages are that it leaves areas on the opposing sides of the adjacent proximal phalanges that require grafts, and that it creates a linear palmar scar at the junction of the web with the palmar skin that may in some cases lead to a secondary contracture across the palmar junction of the web. Upton⁵ proposed a modification that employed a small palmar-based triangular flap to interdigitate with the large dorsal flap. This small flap has two advantages. It breaks up the linear palmar scar and may limit secondary linear contracture, and it adds breadth to the palmar aspect of the web space by expanding the width of the palmar end of the web flap.

In the separation of syndactyly, skin coverage is always deficient (Fig. 44-5). In addition, the use of a large dorsal flap to create the web space leads to tissue deficiency on the dorsal and lateral aspects of adjacent proximal phalanges. The site of the separation of the digits must be resurfaced. We do not agree with the recommendation of several authors that the skin flaps should be preferentially used to resurface one of the two digits, with graft applied to the other.⁴ The design of the skin and subcutaneous flaps should ideally allow the creation of triangular flaps that provide coverage at the level of the PIP joint. Depending on the extent of tissue deficiency, either complete or partial interdigitation is possible. With a skin flap design, this is possible in virtually all cases, with the possible exception of the severe tissue deficiency seen in Apert hands. This approach has two major advantages. First, it provides soft and supple, well-vascularized skin over the joint of the finger that has the highest demands for range of motion. The PIP joint typically has a range of active motion of 135 degrees. The second advantage is that this approach divides the area of full-thickness graft necessary to much smaller areas. Hand surgeons acknowledge that

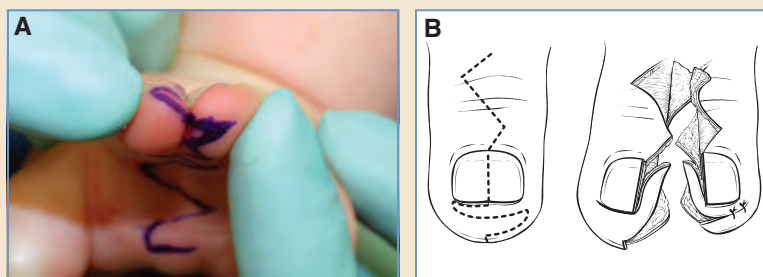


Fig. 44-6 A, Interdigitating flaps are drawn on the hand hyponychium to create digital paronychium after separation of the nail plate (after Buck-Gramcko). B, Interdigitating flaps for reconstruction of the paronychium.



Fig. 44-7 Potential skin graft donor sites for grafting for syndactyly. X marks the anterior superior iliac spine.

any area of graft loss should promptly undergo repeat grafting. This is much easier with smaller areas as opposed to the entire side of a digit.

Separation of the distal phalanges is an important component of separation of digits, and one that requires a well-planned and thorough approach for success. In complete syndactyly, the degree of tissue deficiency guides the approach. Cases of complete syndactyly that do not include a conjoined nail plate have a rim of paronychial tissue. Separation can often be followed by simple advancement and closure of the pulp skin and subcutaneous tissue to this rim of paronychial tissue. If the nail plate is fused, advancement and closure are occasionally possible, but the alternative of interdigitating incisions on the hyponychium should be considered¹⁴ (Fig. 44-6). These incisions create flaps that can be brought proximally to create a functional paronychial reconstruction.

After a web space is created, the distal phalanges are reconstructed, and interdigitating flaps are brought into either alignment or abutment at the proximal phalangeal level. Areas are always

present that require resurfacing with skin grafts. Full-thickness skin grafts are used. These can be harvested from the groin, the medial upper arm, the lower abdomen, or even the foreskin (Fig. 44-7). Areas not to use as donor sites include the palmar wrist and the antecubital fossa, both of which leave considerable unsightly scarring. As Netscher¹² has appropriately pointed out, in children of color, skin grafts tend to hyperpigment, and the dark grafts can be unsightly. Therefore he recommended the use of the light-colored skin from the side of the foot before the child begins to ambulate to provide an appropriate color match. This approach has merit.

Surgical Technique

After anesthesia is given, the upper arm is wrapped with cotton batten or a cotton sleeve, and a tourniquet is placed on the upper arm. Positioning the child's torso on an arm board perpendicular to the main operative table is often helpful. In standard fashion, all intravenous lines, cables, and bipolar cautery are routed away from the end of the hand table so that the end of the hand table is unobstructed. In bilateral cases, a double extremity sheet is useful for draping the child (Fig. 44-8, *A*).

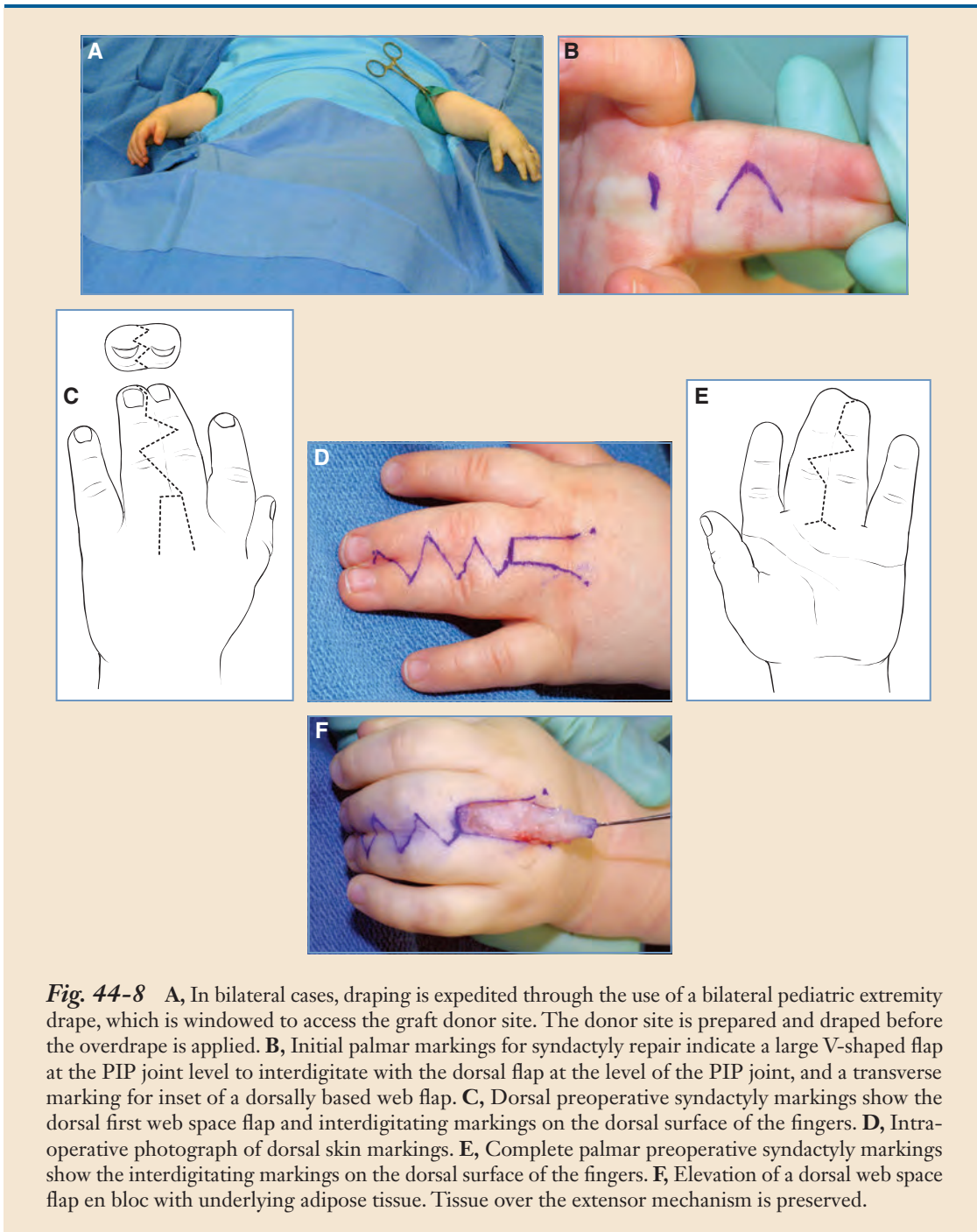
All surgery is performed under loupe magnification. The digits are drawn into flexion, and operative markings are made. The center position of the metacarpal heads is marked on the skin (see Fig. 44-4). The dorsal flap is designed as a trapezoid, extending from the midpoint of the metacarpal heads to a transverse line approximately two thirds to three fourths the distance to the dorsal PIP joint. The flap should be long enough to be transposed to create a new commissure without tension. The level of inset of the flap is then designed on the palm with a transverse line marked at a point 3 to 4 mm proximal to the alignment of the normal cascade of the web spaces (Fig. 44-8, *B*). Normally, the position of the index-long web is at the same level as the long-ring web, whereas the ring-little web is more proximal. This transverse line marking the palmar aspect of the web space is usually placed at a point proximal to the proximal digital crease. Although it is debatable, overcorrection by placing the palmar position of the web space 3 to 4 mm more proximally than the native arcade of the web spaces is reasonable and allows a margin of distal web creep with hand growth. A triangular-based flap can be designed with the base on this transverse line to allow interdigitation with the dorsal flap.⁵

The next step is to plan the closure of the lateral aspects of the digits. A V-shaped flap is designed on the dorsal skin of the hand and a V-shaped flap on the palmar surface of the hand that are based on opposite digits. The apices of the flaps extend approximately halfway across the digit. The adjacent distal flaps are similarly designed to facilitate interdigitation of the flaps and flap approximation at the level of the PIP joint (Fig. 44-8, *C* through *E*).

In complete syndactyly, the design for separating the distal phalanges and the nails is the next step. This uses either advancement of the palmar skin and soft tissue pulp or the interdigitating incisions and flaps developed by Buck-Gramcko¹⁴ (see Fig. 44-6). The dorsal interdigitating flaps between the distal phalanx and the PIP joint are designed, followed by the palmar design of the interdigitating flaps. In this design phase, the priority is to obtain the following:

- Favorable commissure resurfacing
- Optimal resurfacing of the distal phalanges
- Interdigitating flap coverage that reaches across each digit at the level of the PIP joint (see Fig. 44-8, *C* through *E*)

After completion of the flap design, the arm is exsanguinated and the tourniquet inflated to 200 to 225 mm Hg. The incisions surrounding the dorsal flap are made, and the dissection is performed

*Continued*

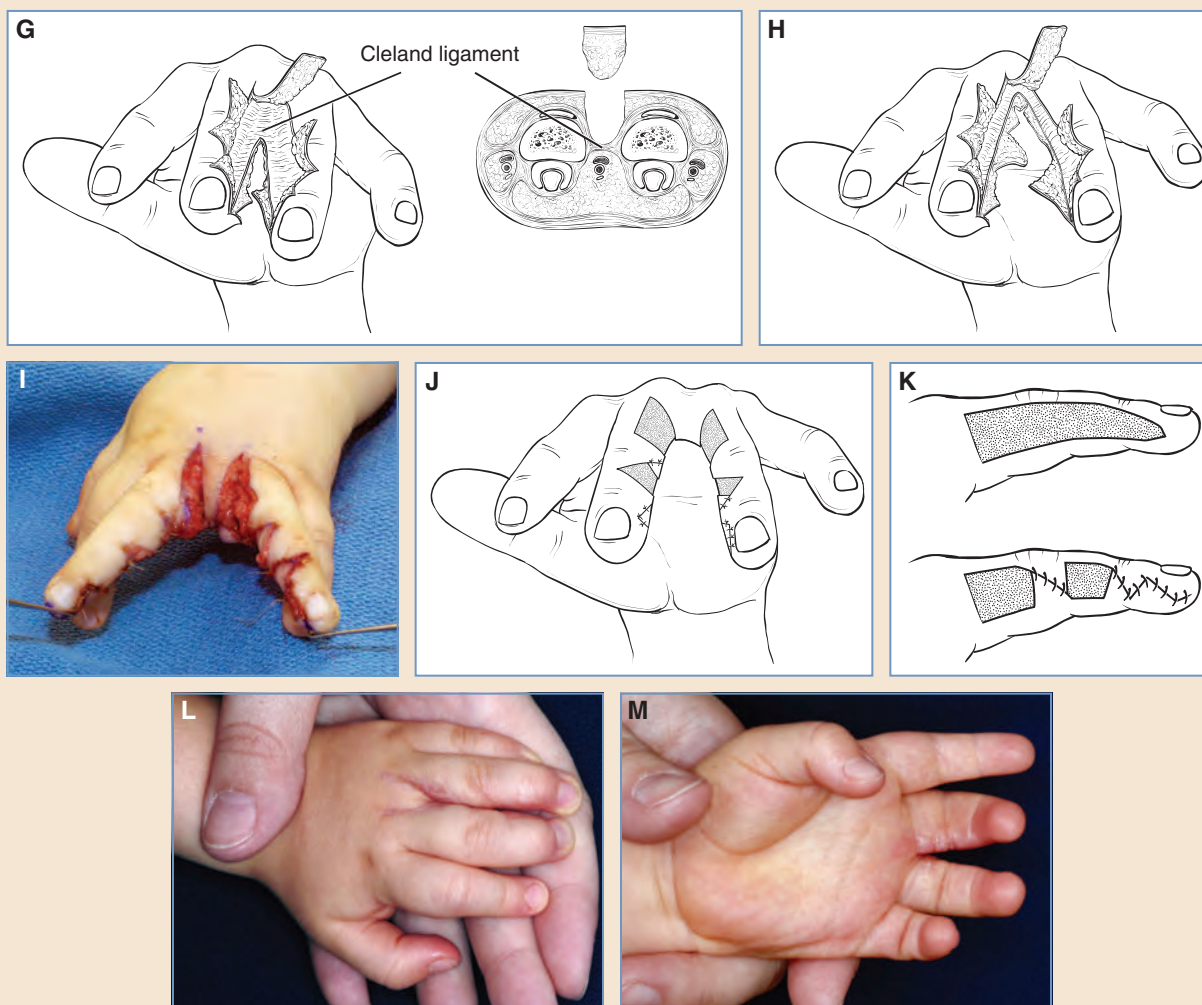


Fig. 44-8, cont'd **G**, Elevation of the dorsal flap reveals conjoined ligaments (consistent with fused Cleland ligaments) on the medial aspects of the digits. Starting distally, these ligaments are carefully divided under direct vision. The neurovascular bundles will lie immediately palmar to the conjoined ligaments, as shown in the cross-sectional diagram on the right, necessitating great care in identifying and preserving these structures. If they are not identified after the division of the first distal portion of the conjoined ligaments, attention is directed proximally, where they should be identified near the web space. The division of this thin layer of conjoined ligaments should proceed from proximal to distal. **H**, Progressive division of conjoined ligaments extends into the level of the web space. Division of the transverse metacarpal ligament is neither necessary nor desirable. **I**, Syndactyly separation after transposition of the web space flap and interdigitating flaps. The intervening areas and the donor site for the dorsally based web flap will require grafting. **J**, The shaded areas are common areas of tissue insufficiency that require grafting. **K**, It is desirable to limit and divide the areas into subunits with flaps (*bottom*) and less desirable to graft large, single areas (*top*). The use of interdigitating flaps over the PIP joint allows the graft to be divided into two smaller areas. **L**, Dorsal view and **M**, palmar view at the nine-month postoperative photographs showing healed syndactyly.

down to the level of the conjoined Cleland ligaments. The dorsal flap is elevated with the subcutaneous fat in the plane directly above the conjoined Cleland ligaments to the level approaching the metacarpal heads (Fig. 44-8, *F*). As the dorsal flap is elevated, care is taken to preserve some tissues over the lateral bands and the dorsal tendon, as well as the venous channels in the flap as much as possible. The veins often become evident as the subcutaneous fat is separated by blunt dissection with tenotomy scissors (see Fig. 44-8, *F*).

Distally, incisions for the flaps to separate the distal phalanges are made. If the interdigitating incisions of Buck-Gramcko¹⁴ are used, they should be made first, preventing injury to the bases of the flaps as the conjoined nail is divided (see Fig. 44-6). Attention is turned to the dorsal surface. The V-shaped flaps are incised and elevated off of the underlying conjoined ligaments (Fig. 44-8, *G*). The conjoined ligaments are divided along the entire length of the syndactyly, from distal to proximal (Fig. 44-8, *H*). The semitranslucent conjoined ligaments can be divided as the neurovascular bundles are identified. If these are not readily identified distally, the conjoined Cleland ligaments are divided at the level of the proximal phalanges, and the neurovascular bundles can be reliably identified at the level in the web and then traced distally to ensure their integrity is preserved. The palmar skin is sequentially incised, verifying the position of the premarked palmar flaps for interdigitation with the dorsal flaps as the division proceeds proximally. The incisions may be manipulated either slightly proximal or slightly distal to maximize coverage at the PIP joint. Next, the web space incision is made approximately 4 to 5 mm proximal to the proximal digital crease, and the dorsal flap is transposed into place and sewn with plain gut suture. The interdigitating flaps are transposed and secured in place (Fig. 44-8, *I*). Thinning the flaps is not usually necessary or desirable; instead, they are moved en bloc with their subcutaneous fat. The flaps are carefully positioned to ensure they are under minimal tension to prevent compromised perfusion. If this is in doubt, the interdigitation of the flaps should be relaxed with less advancement and the surface area to be grafted expanded to promote perfusion.

After transposition of the flaps, a template for assessing the surface area is made based on the size of the defect (Fig. 44-8, *J* and *K*). The tourniquet is released, and the perfusion and viability of the flaps and the digits are assessed. If perfusion and viability are questionable, the interdigitating flaps should be recessed and relaxed with a concomitant expansion of the area to be grafted.

The template is transposed to the donor site. In single digit separations, this is often the groin crease or the lower abdominal wall. In multiple digit separations, the donor site of choice is usually the lower abdominal crease, which has the advantage of providing a large volume of skin with relative ease and minimal tension on closure. Subcutaneous fat is harvested with the graft and defatted on the back table. The graft is thinned to the dermal level with scissors, and perforated or pie-crust by placing it on a folded paper surgical drape and perforating it repeatedly with a No. 15 blade. The donor site is closed in layers and completed with a subcuticular closure and tissue glue. Tissue glue provides an effective barrier while the infant is in diapers.

The perforated graft is tailored and secured under mild tension to the digit using interrupted sutures of plain or mild chromic suture. The interrupted sutures facilitate evacuation of blood from beneath the graft. Running sutures do not allow blood or serum to be evacuated from beneath the graft. Tourniquet time should be minimal (less than 2 hours).

The dressing is very carefully applied, beginning with bridal veil (for example, N-Terface or Dermanet) and a petrolatum ointment. A petrolatum gauze (Vaseline gauze or Xeroform) is then placed as 1-inch-wide strips around 270 degrees of each digit, centered on the site of digital separation and extending onto the adjacent digit. The web space is filled with moistened cotton batten and fluffed cotton gauze. One-inch polyester silk tape is then placed in a noncircumferential fashion, anchoring the web space dressing to the skin of the forearm and upper arm. Alternatively, in isolated cases, if small areas of graft are placed, they can be secured with tie-over bolster dressings using interrupted silk sutures to secure bridal veil, Vaseline gauze, and cotton

batten. In either case, a bulky hand dressing is then applied with fluffs and cotton gauze roll as a wrap. The tips of the digits should be visible at the distal end of the dressing. Three-inch silk tape is then applied to secure the dressing to the proximal forearm and upper arm. A fiberglass or plaster splint is placed to stabilize the hand, forearm, and elbow, with the elbow in 90 degrees of flexion. A stockinette dressing is applied as a swath around the torso. This elbow is carefully wrapped below the contralateral axilla to prevent pressure on the neck. The purpose of this extensive dressing is to secure the grafts to the bed. One manner in which this is achieved is by completely eliminating functional use of the hand, preventing the child from reaching for things. Effective immobilization is crucial to successful graft take and survival (Fig. 44-8, *L* and *M*).

First Web Space

The web space between the thumb and index finger is seldom involved in a true syndactyly in nonsyndromic patients (approximately 5% of cases).⁴ However, the first web space can often be constrained and restrict thumb motion, limiting the span of the hand's grasp and thumb function in congenital disorders. The first web space is critical to the function of the hand and the ability to position the thumb in space, thereby allowing effective grasp. In a normal hand, the thumb accounts for approximately 40% of total hand function (based on amputation at the MP level of the thumb [AMA guide]).¹⁵ In hands with limitations in other digits related to congenital anomalies, the thumb may contribute an even greater component to total hand function. The normal range of motion for the thumb in adults is 50 degrees of radial abduction, 8 cm of opposition (measured from the palmar surface of the thumb to the palmar surface of the base of the long finger in direct opposition), and 8 cm of adduction (the thumb tip to the MP joint of the fifth finger).¹⁵ Adduction contracture is considered significant if the intermetacarpal opening is less than 40 degrees.^{15,16} It is evident that a constrained first web space will affect function in all of these motions and significantly limit hand function. Cases with limited restriction can be treated by instructing the parents in range-of-motion exercises in which stress is isolated to the intermetacarpal space of the first web and performed several times per day. Splinting has not proved particularly beneficial in these children.¹⁶

Release of the first web space varies with the severity of the limitation caused by the restriction. In many cases, the only deficiency is the skin and subcutaneous tissue that creates a tethering effect on the thumb. Several techniques have been described for effective resurfacing of the first web space once a release is performed. All techniques make preferential use of adjacent and remote flaps, as opposed to skin grafts, to resurface the web expansion. The techniques create a gradual U-shaped web, not a steep V-shaped web. For this reason, double Z-plasties are preferred over a single, large Z-plasty. Three primary techniques using Z-plasty are routinely performed. These include a four-flap Z-plasty, a double Z-plasty, and a double-opposing Z-plasty (Fig. 44-9, *A* through *C*). These techniques are the workhorses for expansion and resurfacing of the first web space for moderate expansions. The technique chosen depends on the surgeon's experience and preexisting scars or anomalies that may limit the flap design. Larger expansions of the first web space, which may be necessary for severely constrained first web spaces in Apert syndrome, Poland syndrome, or arthrogryposis, may require dorsal hand rotation flaps.¹⁷ An expansion of the first web space by surgical release should not be compromised by resurfacing with an inadequate surface area of skin or a tight closure.

Additional measures may be needed for cases in which the first web space has a significant and severe adduction contracture. After the skin flap is elevated, the first dorsal interosseous muscle is identified spanning the dorsal web space. The initial step is to release the myofascia investing the first dorsal interosseous muscle. Motion is reassessed, and if the restriction persists, the origin

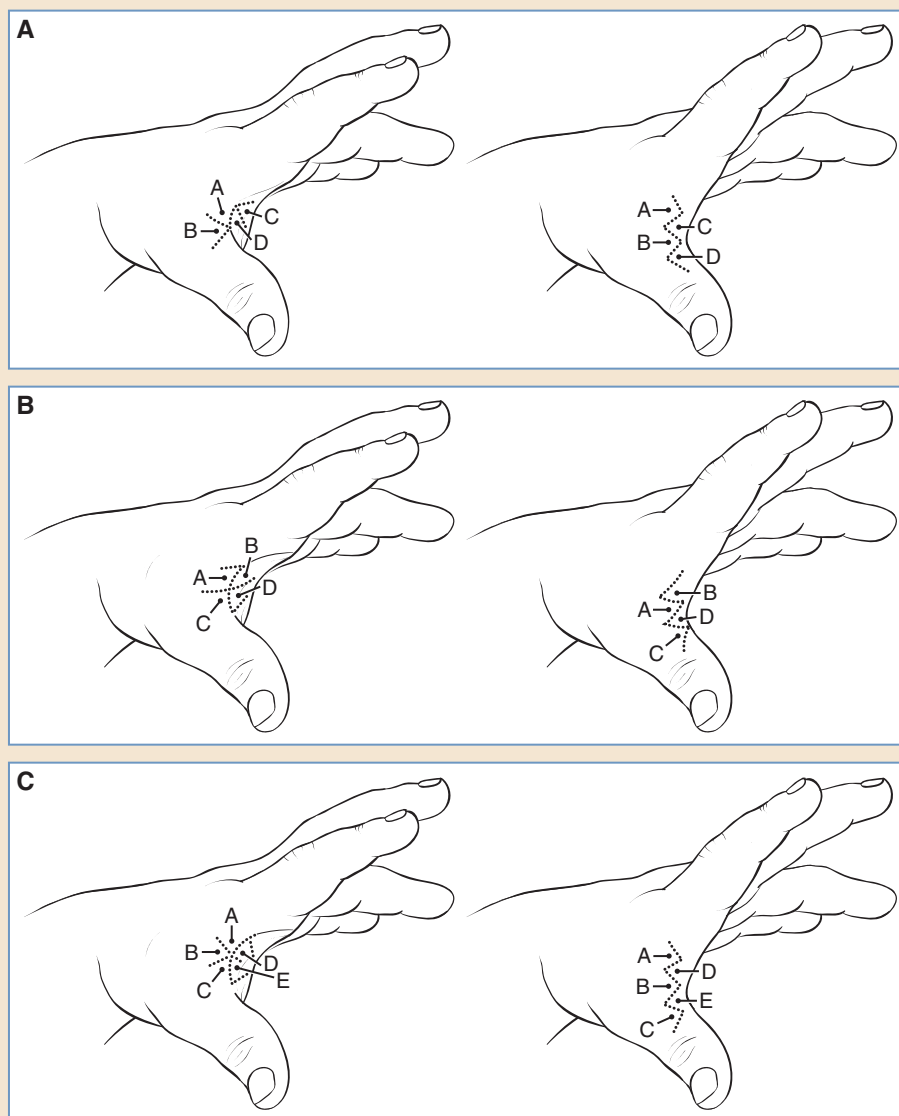


Fig. 44-9 A, Four flap Z-plasty for opening the first web space. B, Double Z-plasty for opening the first web space. C, Double-opposing Z-plasty for opening the first web space.

of the first dorsal interosseous can be partially released from the first metacarpal shaft, and the myofascia investing the adductor pollicis can be released. Great care is required for identifying and preserving the radial digital nerve of the index finger and the ulnar digital nerve of the thumb throughout this portion of the surgery. In severe cases, the insertion of the adductor pollicis can be detached and shifted more proximally on the thumb. The muscle body of the adductor should be protected, because it becomes the primary remaining muscle allowing effective pinch and grasp. Sometimes release of the muscle fascia and/or recession of the muscle or muscles is necessary to

adequately expand the first web space. In these cases, many hand surgeons will place a subcutaneous pin across the web space between the index and thumb metacarpal shafts to maintain the width that has been gained during the healing period to prevent contracture.

Alternative Surgical Approaches to Syndactyly

Although a dorsal-based web flap for commissure reconstruction is perhaps the most frequent approach used in contemporary management of syndactyly, other approaches have been advanced recently. Several of these advocate the use of a dorsal metacarpal flap to line the web space, thereby minimizing or eliminating the need for skin grating. Sherif¹⁸ recommended the use of a V-to-Y advancement dorsal metacarpal flap, and Hutchinson and Frenzen¹⁹ have reported additional experience with a comparable flap. Hsu et al²⁰ have modified this flap design to obtain a more favorable web configuration. Rather than V-Y advancement, Aydin and Ozden²¹ advocated rotating a dorsal metacarpal flap into the web space. These techniques have the advantage of allowing digital resurfacing in select cases without the use of skin grafts, have been shown to be reliable, and can be completed in comparable times; however, they result in a linear dorsal hand scar.

SPECIAL CONSIDERATIONS

Poland Syndrome: Symbrachydactyly

Poland syndrome is an uncommon deformity of the hand, arm, and chest wall that occurs in approximately 1 in every 25,000 births.^{22,23} The most often seen manifestation is a deficiency of the sternal portion of pectoralis major muscle, with preservation of the clavicular head of this muscle.^{24,25} Frequently, maldevelopment of other muscles of the shoulder girdle, deficiency of the subcutaneous tissue, anomalies of the ribs, and breast anomalies are present. In many females, the breasts do not develop at puberty and remain hypoplastic. The entire upper extremity may be slightly hypoplastic. The hand anomaly that typically exists in Poland syndrome is symbrachydactyly—shortened fingers that are fused together. The thumb is frequently involved and may have a constrained first web space. These digits are separated following the principles described previously. However, two important caveats exist. First, the neurovascular bundles frequently divide distally, and this must be anticipated in treatment planning.¹⁴ Second, the timing of separation of the fingers may need to be delayed, because the phalanges may be hypoplastic and the joints unstable. Delaying the separation will allow further growth of the digits and may lead to improved digital stability at the separation. If the digits remain hypoplastic and unstable, the benefits of separation of the syndactyly will be limited.¹⁶ However, the first web space can be released and resurfaced earlier and is not generally subject to this same consideration.

Apert Syndrome: Acrosyndactyly

Apert syndrome is characterized by severe, complex syndactyly involving the hands and a severe craniofacial malformation. It occurs in approximately one of every 150,000 births.²⁶ The disorder has been localized to an anomaly in the fibroblastic growth factor receptor. Unlike other mutations that lead to decreased binding between hormone and receptor, it is thought that the mutation in the fibroblastic growth factor actually leads to an increased binding affinity between the receptor and fibroblastic growth factor, signaling the downstream intracellular effects that lead to Apert syndrome.

Hand involvement in Apert syndrome can be grouped into three separate types:

Type I: *Obstetrician's* or *spade hand*, in which the thumb and fifth finger are largely free

Type II: *Mitten* or *spoon hand*, in which the thumb is typically joined to the hand mass by a simple syndactyly

Type III: *Rosebud* or *hoof hand*, in which the thumb is joined by a tight osseous or cartilaginous union to the finger mass²⁶ (see Fig. 44-1)

All cases typically have a complex union of the index, long, and ring finger distal phalanges. Apert syndrome is a complex anomaly and is discussed in detail in Chapter 45. It is mentioned here, because the syndactyly present in these hands is frequently severe, with significant anomalies of the tendons, nerves, and arteries. Distal division of the arteries and nerves are present routinely. The soft tissue deficiency is profound and may require flap coverage.

Pseudosyndactyly

Constriction Ring Syndrome

Constriction ring syndrome, also known as *amniotic band syndrome*, is a well-known anomaly that affects extremities and the face more frequently than the trunk. The condition is sporadic and not inherited. The overall incidence is approximately 1 in 15,000 births.²⁷ The cause of this disorder has been contentious. Some authors have reported that an intrinsic defect of the mesodermal

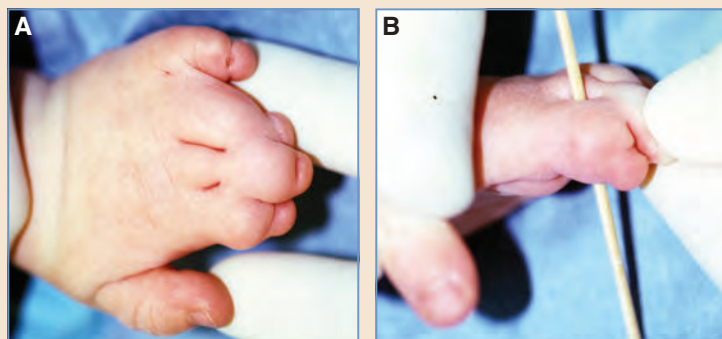
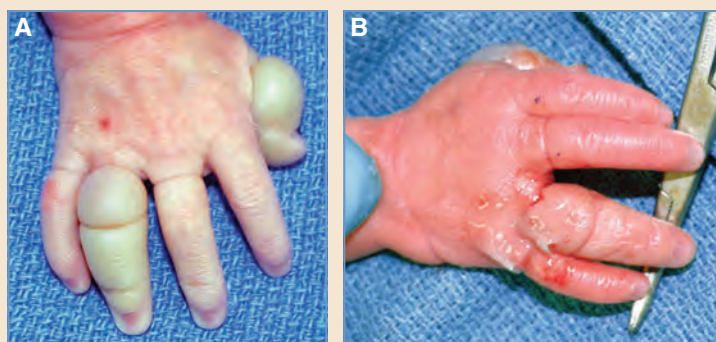


Fig. 44-10 A, Constriction ring syndrome demonstrating distal fusion of the fingers after digital separation. B, Epithelium-lined sinus tracts in constriction ring syndrome.

Fig. 44-11 A, Constriction ring syndrome with ischemic digits in the perinatal period of this premature child. B, A semicircumferential release with Z-plasties was performed under an operating microscope. Perfusion was improved. The second stage of the release was deferred until the child was older (about 6 months of age).



tissue leads to the characteristic constriction rings.²⁸ Today, most authors think that constriction rings are caused by external forces, such as amniotic bands, that encircle and/or compress areas of the fetus and lead to the characteristic deformities. Evidence for this includes a straight line across multiple digits, the presence of a filamentous structure often wound around the structure, and acrosyndactyly in which the digits that have been separated have apparently undergone re-fusion at the site of the band²⁷ (Fig. 44-10, *A*). The hallmark of these hands is the presence of fully lined sinus tracts that pass proximal to the site of distal digital fusion (Fig. 44-10, *B*).

Occasionally, constriction rings lead to ischemia and/or amputation. If this situation is evident at birth, prompt release of the band and Z-plasty is indicated (Fig. 44-11). Separation of the digits involved in constriction ring syndrome is directly analogous to syndactyly separation. Although Buck-Gramcko¹⁴ reported no complications using a single-stage total circumferential release, most surgeons recommend a staged, semicircumferential release. Constriction ring syndrome is discussed in detail in Chapter 49.

Epidermolysis Bullosa

Recessive dystrophic epidermolysis bullosa is an inherited skin disorder manifest by severe blistering disease of the skin and mucosal surfaces in response to mild trauma. The disease is characterized ultrastructurally by cleavage beneath the basal lamina and marked collagen degeneration of the papillary dermis in early blister formation.²⁹ Trauma, even a minor shear stress of the skin, leads to substantial sloughing of the skin. The hand becomes involved because of its position and propensity for trauma. The repeated slough and healing cycles lead to the formation of the regenerating skin that encases the fingers in a pseudosyndactyly, referred to as an *epidermal cocoon*. Treatment requires surgical separation of the digits and grafting, but the disorder is extremely prone to recurrence. Some authors have attempted to use alternative means of tissue coverage, but this experience is limited.³⁰ Care is needed to prevent trauma at all sites, including the oral and pharyngeal mucosa during anesthetics.

Camptodactyly

Camptodactyly, as noted previously, is one of the most common congenital anomalies of the hand.^{4,5} It is a descriptive name for a nontraumatic flexion deformity of the digit, most commonly the little finger; however, multiple fingers can be involved. The typical findings are flexion at the PIP joint with hyperextension of the MP joint.³¹ Most cases arise during the first year of life, but a smaller group of predominantly females will develop camptodactyly during adolescence.³² Approximately two thirds of the cases occur bilaterally.³³ Camptodactyly is often asymptomatic. The disorder is painless. The exact incidence is difficult to establish, because the disorder is probably underreported; however, one author has estimated that it affects less than 1% of the population.³⁴ Most mild cases are ignored by the child and by the parents, but the disorder can be severe, with the development of fixed contractures at the PIP joint, and can even be accompanied by rotatory deformity of the fifth digit into supination.³¹ If left untreated, most cases show some progression, particularly during periods of growth.³¹

Camptodactyly describes the posture or position of the finger and does not indicate a discrete diagnosis or pathogenesis. Many cases of camptodactyly occur sporadically, whereas others are autosomal dominant. Some are related to congenital syndromes, such as trisomy 13, oculodentodigital syndrome, orofacial digital syndrome, Aarskog syndrome, and cerebrohepatorenal syndrome.³¹ The factors possibly contributing to camptodactyly are numerous and varied. Most of the structures in the hand and finger have been suggested as contributors, including aberrant

lumbrical muscles, a band of palmar fascia from the A1 pulley, a band of fibrous tissue in association with the Landsmeer ligament, PIP palmar plate contracture, laxity of the extensor mechanism at the dorsal PIP joint, and anomalies of the flexor tendons. Smith and Kaplan³⁵ have succinctly stated, “Virtually every structure at the base of the finger has been implicated as a deforming factor.” The pathogenetic factors that have been most often identified include an aberrant insertion of the lumbrical muscle and absence or adherence of the flexor digitorum superficialis tendon. McFarlane et al³⁶ have reported anomalous lumbrical muscles in 100% of cases. Kozin et al³¹ noted the same findings and have also elaborated on a classification system for camptodactyly based on the severity of the clinical findings and the reducibility of the PIP joint deformity. The first group includes those in which the PIP joint can undergo complete active extension when the MP joint hyperextension is passively corrected and blocked. In the second group, the PIP joint cannot achieve active full extension while the MP joint is blocked. The third group includes digits with irreducible PIP joints fixed at less than 30 degrees, and the fourth group includes fixed contractures of greater than 30 degrees.

Treatment for the disorder includes operative and nonoperative strategies. Most patients are treated nonoperatively, especially children. Nonoperative treatment includes home therapy and splinting. Home therapy should be performed by the parents or the patient two or three times per day, during which the PIP joint is stretched to correct its flexion deformity. Care must be taken NOT to apply force to the distal interphalangeal joint; otherwise, a secondary hyperextension deformity can result. The therapy should be supplemented with the use of a forearm-based splint worn a minimum of 8 hours each night, or, preferably, 23 hours a day. If the deformity persists after 6 to 12 months of splinting and is severe enough, exploration and correction of the lumbrical and/or flexor tendons is considered. The MP joint can be stabilized by any one of the variety of techniques previously described, including a palmar plate or a Zancolli lasso procedure. Irreducible PIP joints should be splinted. If the deformity is relatively asymptomatic and less than 30 degrees, then splinting is sufficient. If it is more than 30 degrees, then operative management should be considered.

With a wide variety of both causes and patients within each study, the treatment results have varied. Engber and Flatt³² reported that 80% of their cases showed progression of the deformity. Only 20% showed improvement with therapy. In their operative candidates, they report that only one third improved with operative treatment. Siegert et al³⁷ reported that only 18% of their patients with mild-to-moderate contractures (less than 60 degrees) showed improvement with surgery, whereas two thirds improved with nonoperative therapy. In general, most surgeons would not recommend operative intervention for a contracture of less than 30 degrees, and, for contractures of more than 30 degrees, they would consider operative intervention only if a trial of conservative measures had failed.³¹

Failure of Differentiation: Skeletal Tissue

Structural anomalies caused by abnormalities in osseous union occur at all levels of the upper extremity, from the elbow to the distal phalanges. They vary considerably in frequency and in their functional implications. The presence of an anomaly does not indicate that correction is necessary, and the need for surgical treatment is dictated by the functional implications of the anomaly. The anomalies range from irregularities in the shape of a phalanx to synostosis, in which the bones fail to differentiate and become fused into a single structure. Synostosis results from a failure of segmentation of the upper extremity.

Elbow Synostosis

Elbow synostosis can occur either between the humerus and the radius or between the humerus and the ulna. These anomalies are not common. Synostosis at the elbow level can have a profound impact on upper extremity function, with a fusion in extension preventing movement of the hand to the mouth and interfering with feeding, and a fusion in flexion preventing effective reach to the perineum for hygiene.³⁸ A congenital fusion between the humerus and radius can be associated with ulnar ray deficiencies, up to and including absence of the ulna. The synostosis occurs with the forearm and hand positioned in one of two positions: (1) with the elbow in moderate flexion, the radius pronated, and the hand positioned in ulnar deviation, or (2) with the humerus and radius positioned at a near-90-degree angle and the forearm pointing posteriorly. The fundamental principle in these anomalies is that surgery is indicated to improve positioning of the hand, where necessary. This can include rotational osteotomies and must be carefully planned to prevent neurovascular compromise.

Forearm Synostosis

Synostosis of the radius and ulna is usually proximal, but can occur more distally. The extent of the synostosis is variable, and hand function is often not affected.¹⁴ Indications for surgery are controversial and depend on the severity of the deformity and whether the synostosis is present bilaterally.³⁹ Cases with significant pronation deformity may benefit from derotational osteotomy of the forearm. Similar to treatment for elbow synostosis, prevention of neurovascular compromise is essential. The osteotomy is performed through the site of synostosis, and the hand position is corrected. Stabilization is obtained with pins or a plate and screw fixation. Although the derotational osteotomy positions the hand in a more favorable position, there is no establishment of motion of the forearm in the pronation-supination axis; this is something that the child and parents must understand fully before surgery.

Carpal Synostosis

Individual carpal bones separate from a common mass between weeks 4 and 8 of gestation. Incomplete cavitation of the common mass leads to persistent fusion. In contrast to elbow and forearm synostosis, carpal synostosis occurs quite frequently, with an incidence of 0.2% in whites and nearly 9.5% in some West African populations.³⁹ Lunotriquetral synostosis is the most common, followed by fusion between the capitate and hamate.^{14,40} Fusion can also occur between the proximal and distal rows of carpal bones. These fusions are frequently not evident until the second decade of life, when ossification of the carpal bones is complete. Although a congenital synostosis could lead to atypical load patterns within the wrist, most authors report that these anomalies are asymptomatic and result in no wrist dysfunction.⁴¹ Occasionally, a congenital fusion between carpal bones occurs as a synchondrosis with cartilaginous fusion but not as a true osseous fusion. Symptomatic synchondroses have been successfully treated by surgically creating osseous fusion at the affected sites.

Metacarpal Synostosis

Metacarpal synostosis is a rare anomaly that occurs most often between the fourth and fifth metacarpals but occasionally between the thumb and index metacarpal shaft and the central metacarpal shafts.^{14,42} Synostosis of the metacarpals most frequently is seen along the base, but it can extend more distally along the shaft toward the metacarpal head. This congenital anomaly is often associated with syndactyly, central polydactyly, Apert syndrome, symbrachydactyly, and thumb hypoplasia.⁴² When a synostosis occurs between the fourth and fifth metacarpal shafts, the fifth finger frequently can be angulated in an ulnar direction at the MP joint. Indications for surgical treatment depend on the ability to create functional digits after separation.

The fourth and fifth metacarpal fusions are treated by a longitudinal osteotomy extended transversely across the fifth metacarpal base, thereby preventing disruption of the carpometacarpal joint. The MP joint may require reefing and reconstruction of the radial collateral ligaments to correct the abducted posture of the fifth digit. Conjoined tendons need to be separated through the zone of synostosis. Usually, the fifth metacarpal is shortened, and this deficiency is lengthened at the time of osteotomy. The corrected axial metacarpal alignment is then maintained by the use of a block bone graft between the base of the fourth and fifth metacarpal shafts.⁴²

Symphalangism

In 1916 Cushing⁴³ referred to hereditary clinical stiffness of the joints as *sympchalangism*. The name has been extended to describe hereditary stiffness of the IP joints, although it is seldom applied to stiffness of the MP joints. Flatt and Wood⁴⁴ described a subdivision into three types: true symphalangism, symbrachydactyly, and symphalangism with associated anomaly. True symphalangism describes clinically stiff joints with phalanges of normal length. Symphalangism accounted for less than 1% of all congenital anomalies of the upper extremity in their series. It is frequently seen in association with brachydactyly.

Symphalangism is associated with the absence of flexion and extension creases in the affected joint. The absence of digital creases is a necessary but not sufficient criterion for the diagnosis of symphalangism. Stiffness to active and passive motion must also be present. The joint space may be detectable radiologically or clinically. The structural anomaly at the joint can range from partial bridging by cartilage to the complete absence of the joint with the obliteration by bone.

Surgical treatment of this disorder has been largely unrewarding despite multiple approaches, including the microvascular transfer of toe joints.⁴⁵ Deferring treatment until maturity and placing a silicone prosthetic joint have provided the most favorable results.⁴⁶

Clinodactyly

Clinodactyly is angular deviation in the radioulnar plane, whereas camptodactyly is angular deformity in the dorsopalmar plane of the hand. Clinodactyly of the hand typically involves radial deviation of the fifth digit at the level of the middle phalanx. This disorder can be inherited as an autosomal dominant trait with variable penetrance, and, when inherited in this manner, it usually occurs bilaterally.⁴⁷ Clinodactyly can also be inherited in association with multiple syndromes and chromosomal abnormalities.

Clinodactyly is caused by either a trapezoidal- or triangular-shaped bone of the phalanges. This usually exists at the level of the middle phalanx of the fifth digit, but it can involve other

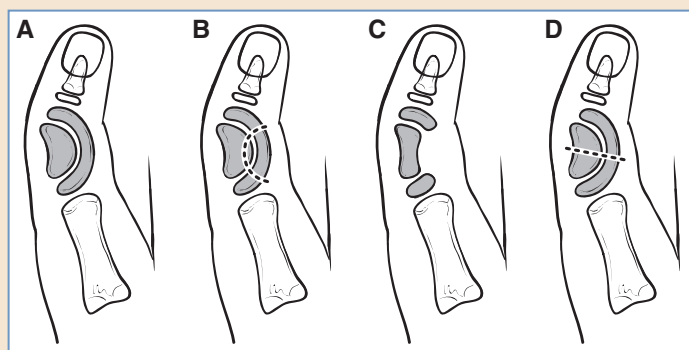


Fig. 44-12 A, Clinodactyly caused by a longitudinally bracketed physis. B, Release of a longitudinally bracketed physis by resection of a portion of the epiphysis and growth plate. C, Release of a longitudinally bracketed physis by resection of a portion of the epiphysis and growth plate, which will be followed by the placement of an interposition fat graft. D, Correction of a longitudinally bracketed physis by a transverse osteotomy combined with either an opening or a closing wedge osteotomy.

metacarpals or phalanges and all rays of the hand, including the thumb. The trapezoidal-shaped bone can have a peculiar growth plate, known as a *longitudinally bracketed physis* or *delta phalanx*⁴⁸ (Fig. 44-12).

Clinodactyly usually does not cause functional impairment. Mild deviation (deviation less than 10 to 15 degrees) of the digits in the radioulnar plane may even be considered within the range of normal. When a deformity is more severe and leads to either a significant cosmetic deformity or functional difficulty, treatment can be beneficial. In these severe cases, splinting is ineffective, and a surgical osteotomy is often required. In cases with a delta phalanx (or longitudinally bracketed epiphysis), a surgical osteotomy can be performed through the abnormal physal plate. A segment of the epiphysis and growth plate is resected (bracket resection) (see Fig. 44-12, C), and an interposition graft of fat or other material is placed in the gap to prevent recurrence.⁵ Alternatively, the digit can be treated using an opening or closing wedge osteotomy⁵ (see Fig. 44-12, D). In cases amenable to bracket resection, surgery is effective at an early age but can be beneficial at any time before fusion of the physis. A formal opening or closing wedge osteotomy is technically challenging, particularly in a child's hand, and this should be considered in surgical planning. In patients who require a wedge osteotomy, surgery should be deferred until the child is older (8 to 12 years of age) and the bones large enough to allow effective control and surgery.

Triphalangeal Thumb

A triphalangeal thumb is often described as a *hypersegmentation abnormality* in which an additional phalanx develops during embryogenesis and differentiation. The additional phalanx may have the appearance of a triangular or delta phalanx, a rectangular phalanx, or a fully formed third phalanx⁴⁹ (Fig. 44-13). It is frequently associated with other anomalies, including polydactyly of the thumb, cleft feet, blood dyscrasias, and congenital heart disease. The Holt-Oram syndrome⁵⁰ is particularly common, and associated anomalies include atrial septal defect, ventricular septal defect, transposition of great vessels, and anomalous coronary arteries.⁵¹ The thumb anomaly itself has essentially three main problems: an abnormal phalanx, a first web space contracture, and hypoplasia of the thenar musculature.⁴⁹ Although function with a triphalangeal thumb may be adequate, the digit itself looks malformed, either from excessive length or from angular deviation caused by the third phalanx. The treatment for this disorder is surgical correction, and most surgeons recommend that it be completed during the first year of life.^{12,49} In surgical removal of



Fig. 44-13 Radiograph of a triphalangeal thumb.

the third phalanx, the periosteum and collateral ligaments both proximally and distally are preserved as much as possible. These are then sewn together, and the thumb is immobilized with a K-wire for 4 to 6 weeks. The deficiency of soft tissue in the first web space is corrected using one of the techniques described previously for syndactyly of the first web space. These include combinations of Z-plasties and the use of a dorsal hand rotation flap, depending on the degree of soft tissue needed. Thenar hypoplasia can be corrected at the initial surgery, or, if this is an equivocal finding, it can be observed and treated later as indicated. Options for treatment include various opponensplasties, such as a Huber transfer, an flexor digitorum superficialis transfer, or a palmaris longus transfer.

Congenital Tumorous Conditions

VASCULAR SYSTEM LESIONS

Vascular lesions of the upper extremity include hemangiomas and vascular malformations and share many similarities with their presentation in other regions of the body. Vascular tumors are the most common tumors of childhood.⁵² Almost invariably, these are benign lesions, with malignant vascular tumors being very rare and accounting for less than 2% of reported series.^{52,53} Mulliken and Young's text and classification system⁵⁴ provides a valuable framework for understanding this diverse spectrum of lesions.⁵⁵ Vascular malformations of the upper extremity are presented in detail in Chapters 11 and 13.

NERVOUS SYSTEM LESIONS

Neurofibroma or Neurofibromatosis

In children, neurofibromas may exist as isolated entities or as a part of the genetic disorder of neurofibromatosis type 1 (NF-1). NF-1 is common, with an estimated 100,000 cases in the United States alone. The clinical hallmark of the disorder is the development of multiple cutaneous and subcutaneous nodular tumors. The disease has a variable age of onset, a variable presentation,

Box 44-3 Clinical Diagnostic Criteria for Neurofibromatosis**Neurofibromatosis-1 (NF-1)**

Diagnostic criteria are met in an individual if two or more of the following are found:

- Café-au-lait spots (six or more larger than 5 mm in greatest dimension in prepubertal patients and larger than 15 mm in postpubertal patients)
- Two or more neurofibromas of any type or one plexiform neurofibroma
- Freckling in the axillary or inguinal region
- Optic glioma
- Two or more Lisch nodules (hamartomas of the iris)
- A distinctive osseous lesion such as sphenoid wing dysplasia or thinning of the long bone cortex with or without pseudarthrosis
- A first-degree relative (parent, sibling, or offspring) with NF-1 by the above criteria

Neurofibromatosis-2 (NF-2)

Diagnostic criteria are met if a patient has the following:

- Bilateral eighth nerve masses seen with appropriate imaging techniques
- A first-degree relative with NF-2 and either:
 - Unilateral eighth nerve mass or
 - Two or more of the following
 - Traumatic injury
 - Neurofibroma
 - Meningioma
 - Glioma
 - Schwannoma
 - Juvenile posterior subcapsular lenticular opacity

Data from 1987 National Institutes of Health Consensus Conference.

variability in clinical findings, and a variable but progressive course. Over the past 30 years, our understanding of neurofibromatosis has advanced significantly. Critical to this advance was a National Institutes of Health Consensus Statement in 1987^{56,57} that established the diagnostic criteria for peripheral neurofibromatosis, now known as *neurofibromatosis-1*, and central neurofibromatosis, now known as *neurofibromatosis-2* (NF-2) (Box 44-3). The prevalence of NF-1 is approximately 1 in 3000,⁵⁸ and the prevalence of NF-2 is approximately 1 in 60,000.⁵⁹ Both disorders are transmitted in an autosomal dominant fashion and associated with variable penetrance and expressivity. Families should be counseled that the chance of having an affected child is 50%. The gene for NF-1 has been localized to band 11.2 of the long arm of chromosome 17. The gene for NF-2 has been localized to the middle of the long arm of human chromosome 22, which is clearly distinct from the gene for NF-1.

Although the genes for both NF-1 and NF-2 have been identified and localized, the degree of variability is so high at these gene sites that the diagnosis of this disorder remains based on the clinical criteria.⁶⁰ The clinical presentation of NF-1 is most often heralded by the appearance of café-au-lait spots. These lesions are cutaneous hyperpigmented areas, typically 20 to 30 mm in diameter, and are the most common manifestation of NF-1, with more than six lesions found in 90% to 99% of all cases.^{61,62} Most children present with café-au-lait spots as the earliest manifestation of NF-1, but more than 80% will develop additional signs of the disorder. Axillary and



Fig. 44-14 An isolated neurofibroma of the distal radial sensory nerve in a patient with NF-1.

skin-fold freckling is seen in approximately 80% of all cases of NF-1.⁶¹ Freckles are generally extremely common, although not in areas that have not been exposed to the sun. Lisch nodules are pigmented, dome-shaped nodules on the surface of the iris.^{61,62} They appear later in life, usually by 10 years of age, and are present in nearly all NF-1 patients by 20 years of age. Lisch nodules are melanocytic hamartomas that are noteworthy for their diagnostic relevance, but they do not require treatment.

The NF-1 gene is a tumor suppressor gene. Optic pathway gliomas are the most common central nervous system tumors in patients with NF-1, occurring in approximately 15% of cases, and are histologically identified as low-grade pilocytic astrocytomas.^{61,62} NF-1 patients also have an increased incidence of brainstem gliomas and an apparent increase in the occurrence of benign and malignant astrocytomas, ependymomas, meningiomas, medulloblastomas, and malignant schwannomas. Skeletal abnormalities associated with NF-1 include sphenoid wing dysplasia, macrocephaly, scoliosis, and thinning of long bone cortex, frequently manifest as anterior tibial bowing.

Neurofibromas, the hallmark of the NF-1 disease, are nerve sheath tumors that may arise anywhere along a nerve sheath from the dorsal root ganglion to the terminal nerve branches⁶¹⁻⁶³ (Fig. 44-14). They are composed of Schwann cells, fibroblasts, mast cells, and perineural cells. The natural history of the disorder is characterized by steady growth of the neurofibroma, starting in childhood.⁶⁴ Surgical resection is the mainstay of treatment for enlarging or symptomatic tumors.

Neurofibromas occur in five main types: localized cutaneous neurofibromas, diffuse cutaneous neurofibromas, localized intraneural neurofibroma, plexiform neurofibromas, and massive soft tissue neurofibroma.⁶³ Localized cutaneous neurofibromas are the most common form. They are slow growing, and typically occur as multiple lesions that progressively increase in prominence throughout life. These lesions cause the pedunculated skin growths so frequently seen in NF-1. Diffuse cutaneous neurofibromas are plaquelike thickenings of the dermis and subcutaneous tissue that are most frequently found in the head and neck regions. They are nondestructive and grow along fibrous septa in children and young adults. They are generally soft and compressible to palpation. Localized and diffuse neurofibromas are generally absent at birth, but may appear as early as 5 years of age. Localized intraneural fibroma is the second most frequent type of neurofibroma and is the fusiform enlargement of a peripheral nerve. These are the most common type of neurofibroma of the upper extremity, accounting for 85% of cases.⁶⁵ Both spinal and cranial

nerves can be involved anywhere from the root to the smallest branches. Massive soft neurofibromas, previously known as *elephantiasis neurofibromatosa*, are rare in NF-1 and rare in the upper extremity. Plexiform neurofibromas are virtually unique to NF-1.⁶³ A plexiform neurofibroma outside NF-1 is extremely rare. They are composed of nerve sheath cells that proliferate along the length of a nerve, and are frequently associated with hypertrophy of the soft tissue and hyperpigmentation or hypertrichosis of the overlying skin. Their growth can cause destruction or compression of local tissue, leading to significant morbidity. Plexiforms occur in 16% to 40% of patients with NF-1. They are found on the trunk in 43% to 44% of patients, on the extremities in 15% to 38%, and on the head and neck in 18% to 42%.⁶³ In contrast to other neurofibromas, these plexiform lesions are thought to be congenital in origin and usually become evident by 2 years of age, perhaps only as an area of pigmentation or subtle soft tissue enlargement. Their growth is unpredictable and often occurs during early infancy and times of hormonal change such as preadolescence, adolescence, and pregnancy.

Malignant degeneration of peripheral nerve sheath tumors is more common than most think, occurring in up to 13% of patients with NF-1.⁶¹ Malignant peripheral nerve sheath tumors, which were formerly known as *neurosarcomas* or *malignant schwannomas*, arise from Schwann cells. Malignant soft tissue neoplasms occur approximately 34 times more frequently than in a control group and account for 9.4% of the deaths of patients with NF-1.^{61,63} More than 50% of patients with malignant nerve sheath tumors have NF-1.⁶³ Only the plexiform neurofibromas have a high propensity for malignant degeneration. Medium and large nerves, such as those involving the thigh, the buttock, the brachial plexus, and the paraspinal nerves, are usually involved. Pain appears to be the most reliable indicator of malignant degeneration. Prompt medical attention is warranted, and surgical biopsy is indicated. Once diagnosed, management consists of an aggressive attempt at total surgical resection. Metastases are common. Regardless of treatment, the 5-year survival rate of malignant peripheral nerve sheath tumors is estimated to be 16% to 52%.⁶³

Despite the possibility of malignant degeneration, surgical management of neurofibroma and neurofibromatosis in the child is usually based on balancing the benefit of tumor debulking/resection versus the potential loss of sensibility and/or motor function. Fundamental principles of surgical technique include the isolation of the peripheral nerve proximal and distal to the area involved; however, it is rarely possible to dissect through the neurofibroma and ensure the integrity of the nerve bundles. Debulking/resection is a surgical reduction in volume and may have consequences distal to the site of resection. The benefit of volume reduction and the potential improvement in pain must be weighed against the possible distal effects of major peripheral nerve surgery. In cases in which the continuity of nerves is interrupted, nerve grafting should be performed.

CONNECTIVE TISSUE LESIONS

Juvenile Aggressive Fibromatosis or Desmoid Tumor

Aggressive fibromatosis arises from connective tissue or muscle. The average age of onset of aggressive fibromatosis is the third decade of life, although it can develop as early as 1 month of age.⁶⁶ Approximately 5% of lesions are found in the hand. The lesion presents as a mass. Pain is seldom present. The tumor is benign but acts aggressively, with recurrence rates as high as 73%.⁶⁶ Recurrence with negative margins is low, but it is as high as 90% with positive margins and can occur within 3 months of excision.⁶⁶

Infantile Digital Fibromatosis or Reye Tumor

Infantile digital fibromatosis occurs as a single nodule or multiple lesions on the fingers and toes.⁶⁷ The lesions nearly always arise on the digit and typically present as a dome-shaped, smooth lesion on the extensor surface. In 80% of patients, the lesions are diagnosed during the first year of life.⁶⁸ Conservative treatment is indicated, but if functional problems or deformities arise, surgery is indicated. Surgical excision is the treatment of choice, but lesions recur in approximately 60% of patients, and this is the reason for the name *recurrent infantile digital fibroma*.⁶⁸ The lesions must be completely excised to prevent recurrence. Those that recur should be re-excised in a nonmutilating manner and grafted or covered with a pedicled flap, as indicated.

SKELETAL TISSUE LESIONS

Osteochondromatosis

Osteochondroma is the most common benign tumor of bone. It is generally asymptomatic, but it may be noticed because of its prominence or its impingement on other structures. It is extremely uncommon in the hand. The lesions are osseous bone growths with a hyaline cartilaginous cap. They can be associated with multiple hereditary exostoses, and these patients have a distinct skeletal problem—a lack of tubulation of bone.⁶⁹

Enchondromatosis

Enchondromas are the most common tumor of bone in the hand. They are benign cartilaginous lesions that typically arise in the proximal phalanx. In contrast, multiple enchondromatosis, or Ollier disease, is an uncommon nonhereditary skeletal disorder that is characterized by multiple prominent growths that cause obvious deformity and can impair function. The lesions usually continue to grow until the time of skeletal maturity and are generally not painful. They have a significant risk of malignant degeneration (approximately 30%), and any onset of pain, continued growth after maturity, or radiographic progression.^{70,71} Patients need to be followed clinically and radiographically for evidence of change that might suggest degeneration; a biopsy is performed if necessary.

Mafucci syndrome is characterized by multiple enchondromatosis, similar to Ollier disease. However, these patients also have multiple hemangiomas. The disorder is rare. The most common site of involvement is the hands. The radiographic appearance is identical to that of Ollier disease and has a similar potential for malignant degeneration. These patients must be followed clinically and radiographically.

Fibrous Dysplasia

Fibrous dysplasia is a benign disorder of bone that affects both the axial and the craniomaxillofacial skeleton. Hand involvement is rare. Fibrous dysplasia has not been classically considered a congenital disorder, because it is not usually evident at birth, but becomes clinically evident during late childhood or adolescence. Most importantly, it occurs sporadically, and genetic transmission has not been documented. Fibrous dysplasia has been traditionally divided into three main categories: (1) monostotic (or monocystic) fibrous dysplasia, (2) polyostotic fibrous dysplasia, and (3) the McCune-Albright syndrome. Most patients (70% to 80%) present with a single

area of bony involvement (monostotic or monocystic fibrous dysplasia); whereas others present with multiple sites of bone involvement (polyostotic fibrous dysplasia).⁷² Histologically, fibrous dysplasia is characterized by an expansile process with an accumulation of disconnected trabecular, poorly mineralized bone matrix mixed with normal lamellar bone. The histologic processes include marrow invasion, hyperresorption of bone with the development of lytic areas, and immature bone formation. The marrow cavity is filled with preosteoblastic cells. In the past decade, it was established that fibrous dysplasia results from a postfertilization somatic cell line mutation leading to a structural and functional change in the cellular transduction mechanism involving G-proteins.⁷²⁻⁷⁴ The G-protein is a membrane bound intracellular signaling mechanism that carries the message of extracellular hormone binding into the cell to create an effect. Hand lesions may remain asymptomatic. If fracture occurs from decreased bone strength, healing is usually uncomplicated in the hand.

CONCLUSION

Failure of differentiation of the hand includes a broad spectrum of disorders involving skin, connective tissue, muscle, nervous tissue, and vascular tissue. The disorders range from those that are very frequent (syndactyly, polydactyly, and camptodactyly) to those that are rare. Familiarity with the disorders in this group of anomalies is essential for providing care for these children. Each common disorder has several core principles.

KEY POINTS

Congenital Flexion Deformities of the Hand

- Assessment of a thumb flexion disorder requires evaluation of associated clinical findings and may include a variant of normal development, a congenital trigger thumb, or a congenital clasped thumb (supple or complex).
- Differentiation between these disorders is based on the joint that is flexed: IP joint flexion is probably a trigger thumb, whereas MP joint flexion can be a normal variant or a congenital clasped thumb.
- The initial management of the disorders with flexion at the MP joint is splinting, followed by reassessment every 2 to 3 months.

Syndactyly

- Syndactyly is one of the three most common congenital hand anomalies.
- A common classification system describes deformity as simple or complex and complete or incomplete.

- Surgery is typically performed during the first year of life.
- In hands with multiple digital involvement, only one side of a digit should be operated on in an operative procedure to prevent a vascular catastrophe.
- All syndactylous digits have a skin deficiency.
- Six fundamental tenets should be followed:
 1. All osseous and soft tissue between afflicted digits at phalangeal levels is completely separated.
 2. Nerves and the vascular supply are optimally preserved.
 3. An adequate web space is developed between the digits.
 4. Interdigitating flap coverage is ensured over the mid-lateral digit.
 5. Appropriate distal phalanges are developed.
 6. Full-thickness perforated grafts are used for areas of tissue deficiency.
- Effective immobilization of the upper extremity is essential to optimize graft adherence and clinical results.

Camptodactyly

- Camptodactyly is characterized by a flexion deformity of the affected digit in the dorsopalmar plane.
- This is one of the most common congenital anomalies.
- It is often associated with an absent lumbrical muscle.
- Splinting has limited effectiveness as treatment.
- Surgical correction is usually limited to digits with greater than 30 degrees of flexion.
- Surgical intervention has limited success.

Symphalangism

- Stiffness of the joints is associated with the absence of flexion and extension creases of the affected digits.
- Treatment is not usually beneficial.

Clinodactyly

- Clinodactyly is an angular deformity of the digit in the radioulnar axis.
- It is often caused by a malformed phalanx: a trapezoidal or a true delta phalanx.
- Often, this condition does not cause a functional impairment.
- Clinodactyly usually involves a mild deviation of 10 to 15 degrees from normal.

Continued

KEY POINTS (continued)

Triphalangeal Thumb

- Triphalangeal thumb is frequently associated with other congenital anomalies, especially cardiac.
- It involves the following separate problems: an abnormal phalanx, a constrained first web space, and hypoplasia of the thenar musculature.
- Many surgeons favor resection of the abnormal phalanx during the patient's first year of life.

Neurofibroma and Neurofibromatosis

- Neurofibromas exist in five main types.
- If a neurofibroma is diagnosed, the possibility of neurofibromatosis must be evaluated.
- If resection is required because of symptoms, the integrity of nerve tracts should be maintained, if possible, and those that are affected should be grafted.

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45

The Apert Hand

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Complex Surgical Case: Postoperative Outcome; Complex Surgical Case: Surgical Planning and Outline of Incision.



Apert syndrome is one of the craniosynostosis syndromes and is associated with symmetrical and severe bilateral syndactylies of the hands and feet (Fig. 45-1). The syndrome's many craniofacial anomalies include craniosynostosis, midface hypoplasia, and ocular hypertelorism with proptosis. Other malformations of the skin, the skeleton, the brain, and other internal organs occur with variable frequency. Apert¹ first described this syndrome in 1906.

Hand function in affected patients is limited by the presence of bilateral, symmetrical complex and complicated syndactylies with symphalangism, and, in some cases, metacarpal synostosis.

The entire upper limb may be affected. When synostosis involves the shoulder and elbow, the hand anomalies are usually less severe. Less than 5% of Apert children have congenital shoulder synostosis and rely on scapulothoracic movement.² However, most have a small glenoid fossa.

With growth, a discrepancy in size between an abnormally shaped humeral head and the glenoid fossa develops, although limitation of movement may not reveal itself fully until the patient has skeletal maturity. Shoulder range of motion tends to decline over time. For this reason, an orthopedic consultation is warranted. The elbow functions normally in most Apert patients, but 50% of patients show some radiologic anomalies.²



Fig. 45-1 A, Children with a typical Apert appearance. B, Typical Apert hands. C, Typical syndactyly of the toes.

GENETICS

The birth prevalence of Apert syndrome is thought to be approximately 1 per 65,000 live births (15.5 per 1 million live births). This constitutes approximately 4.5% of all craniosynostosis cases.³ Most cases occur as a result of new mutations—with a paternal age effect—and rare instances of inheritance are consistent with an autosomal dominant pattern.⁴ This condition is one of five craniosynostosis syndromes associated with allelic mutations of the fibroblast growth factor receptor 2 gene (*FGFR2*).

Nearly all known cases are accounted for by new mutations in the *FGFR2* gene. These mutations are distinct in position and nature.⁵ FGFRs are transmembrane receptor tyrosine kinase proteins that are involved in signaling pathways by interaction with fibroblast growth factors. They are strongly implicated in limb bud development anomalies.⁶

Two particular activating mutations are responsible for the changes seen. These cause amino acid substitutions: Ser 252 → Trp and Pro 253 → Arg (serine 252 to tryptophan and proline 253 to arginine).

Slaney et al⁷ analyzed the DNA of a group of patients and found a significant association between a particular mutation and the presence of either cleft palate (associated with a more severe craniofacial phenotype) or severity of syndactyly. The hand anomalies were most severe in those with the Pro 253 → Arg substitution, revealing a subtle but significant phenotype difference associated with each specific mutation.

ANATOMY

Anatomy of Abnormal Findings in the Apert Hand

The hands in Apert children are strikingly symmetrical. The deformity is complex, and the more common forms present with the following:

- Complex syndactyly of the index, middle, and ring fingers
- Simple syndactyly of the ring–little finger web
- Symphalangism of the index, middle, and ring fingers
- A short thumb with radial clinodactyly

Skeletal Abnormalities

Skeletal abnormalities include the following (Fig. 45-2):

- Capitohamate coalition at the carpal level
- Fourth to fifth finger metacarpal synostosis
- A bracketed longitudinal epiphysis on the radial border of the thumb proximal phalanx
- A broad and radially deviated thumb
- Stiff digits from symphalangism of the proximal and middle phalanges

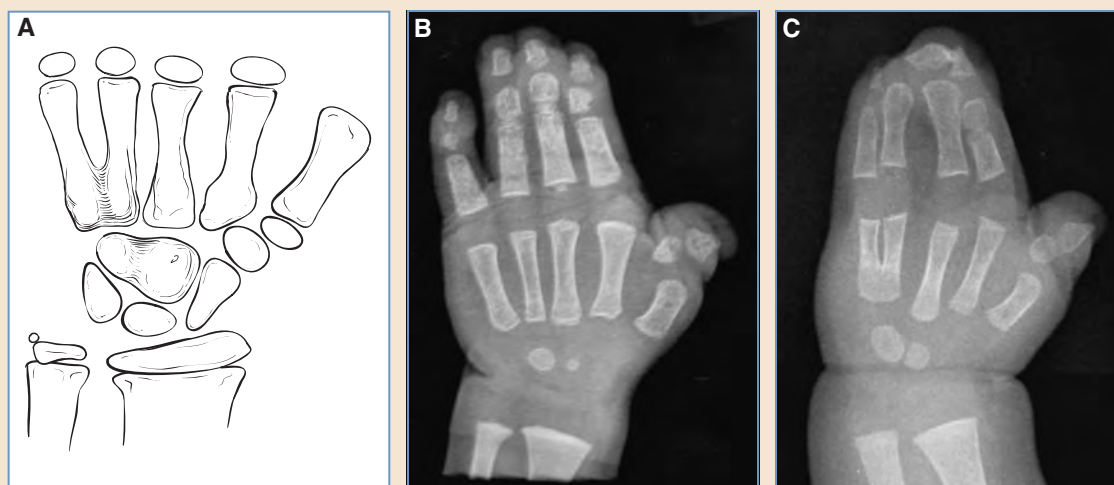


Fig. 45-2 A, A left hand with fourth to fifth metacarpal synostosis and capitohamate coalition. B, Radial deviation of the thumb, resulting from a delta phalanx. C, Symphalangism with no proximal interphalangeal joint visible in the third, fourth, and fifth fingers.

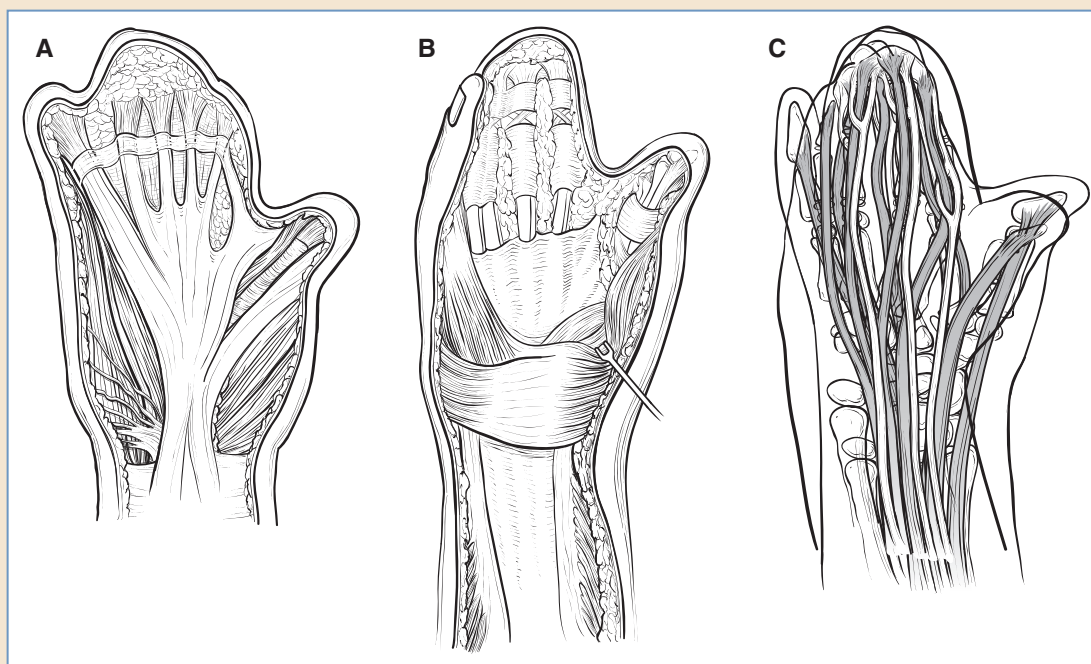


Fig. 45-3 A, The palmar aponeurosis extends to the radial border of the thumb. B, The digital sheath is intact, with cruciate portions present only at areas of joint motion. C, The superficialis tendons arborize in a distal position and sometimes encircle the profundus tendon.

Soft Tissue Abnormalities

Large bands extend from the palmar aponeurosis to the radial border of the thumb. Strong interdigital bands connect the phalanges of the first web. The thenar muscles are always present, and the adductor pollicis and first dorsal interosseous muscles are often large when observed at the time of first web release.

A continuous digital retinacular sheath with cruciate portions is only present where joint movement is observed. The common digital neurovascular bundles tend to arborize in a distal position in each web. Bifurcations of the superficialis tendons are distally positioned within each digit.

Lumbricals are present, but with abnormal distal insertions into a variety of coalesced phalanges. They contribute to metacarpophalangeal flexion but not to interphalangeal extension⁸ (Fig. 45-3).

CLASSIFICATION

Syndactyly of the first web space occurs variably. Upton^{8,9} used this as a basis for classification of the Apert hand into three types. Type I has a spadelike appearance; type II is spoonlike or mittenlike; and type III is hoof or rosebud shaped. This classification is in general use. However, Van Heest et al¹⁰ described a classification based retrospectively on the radiographic features of the presenting anomalies of each hand and the surgical treatment provided.

Functional Implications

The severity of functional difficulty increases with classification type, beginning with type I. A lack of span in the first web prevents adequate grasp of larger objects, and syndactyly of the other digits limits a patient's ability to perform fine movements. Individual phalangeal joint movements are limited; therefore the digits are stiff, and finger flexion occurs at the level of the metacarpophalangeal joints and the distal interphalangeal joint of the little fingers. Radial deviation of the thumb impedes the pincer grip.

Cosmetic Implications

Even the mildest form of Apert syndrome hand has an unusual and abnormal appearance, compounded by the functional limitations of the condition. The hands are the next most visible area after the face and head and warrant consideration for treatment.

Descriptions of Classification Types

Type I

The type I hand, formerly described as *spade* or *obstetrician's hand*, is the least severe and the most common type (Fig. 45-4). The thumb-index web syndactyly is simple and relatively mild; thus

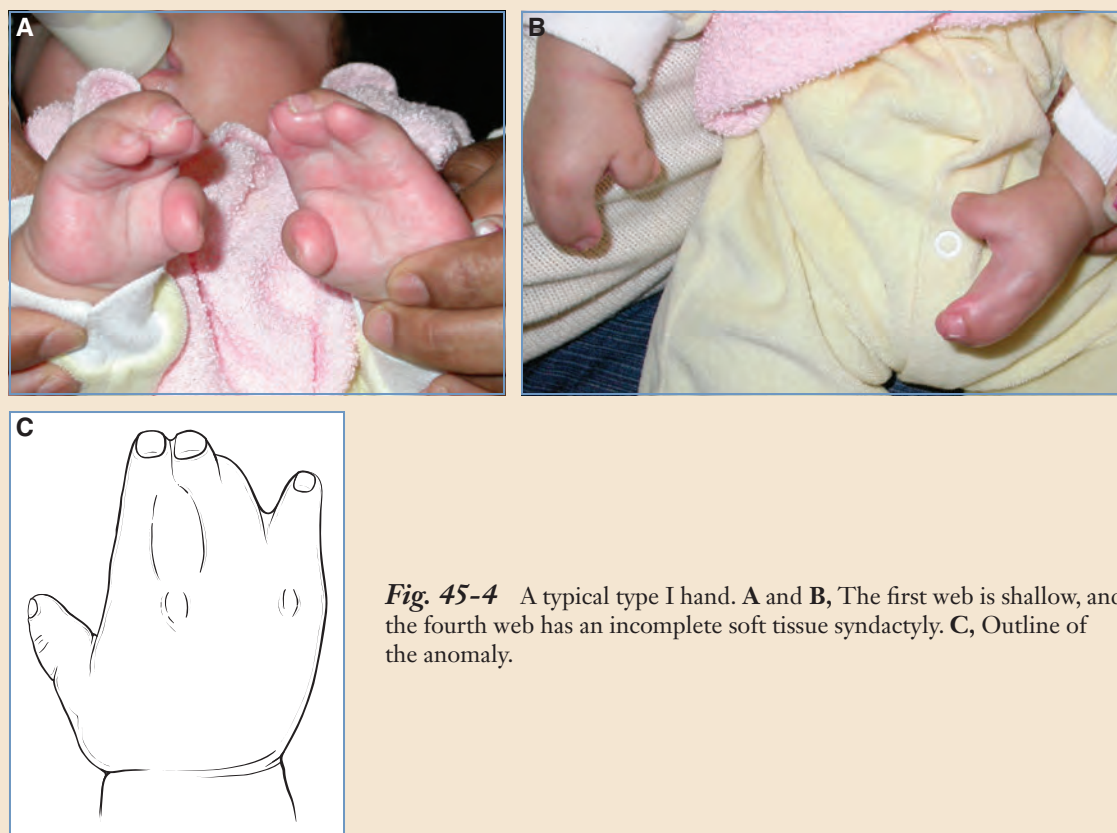


Fig. 45-4 A typical type I hand. **A** and **B**, The first web is shallow, and the fourth web has an incomplete soft tissue syndactyly. **C**, Outline of the anomaly.

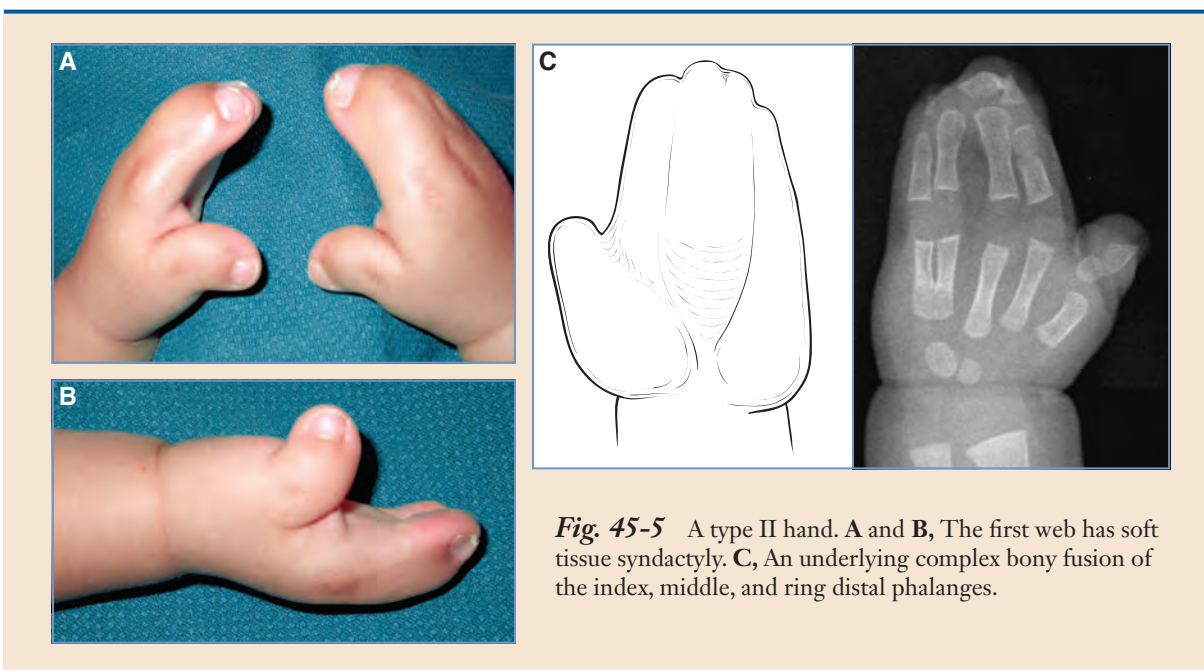


Fig. 45-5 A type II hand. **A** and **B**, The first web has soft tissue syndactyly. **C**, An underlying complex bony fusion of the index, middle, and ring distal phalanges.

the first web space is shallow. A variable degree of radial clinodactyly may be present. The index, middle, and ring fingers are fused. A simple syndactyly occurs in the fourth web. The index, middle, and ring fingers have a bony fusion at the level of the distal interphalangeal joint, the distal middle phalanx, or the proximal terminal phalanx. Some degree of movement of the distal interphalangeal joint of the little finger is always present, unlike the other finger joints. Almost a full range of movement is possible at all of the metacarpophalangeal joints. The fused fingers form a flat configuration, and the thumb can oppose well.

Type II

The type II Apert hand was formerly described as having a spoonlike or mittenlike appearance (Fig. 45-5). Type II deformities are more severe than type I, because the thumb is joined to the rest of the fingers by a soft tissue syndactyly. However, there is no bony union, and the thumb and index finger have separate nails. The spoon-shaped appearance is caused by the complex fusion of the distal phalanges of the index, middle, and ring fingers and associated splaying of the proximal phalanges and metacarpals. The palm has a concave appearance. The syndactyly of the fourth web is complete, and there is a single conjoined nail for the index, middle, and ring fingers with some longitudinal ridges representing the individual distal phalanges underneath.

Type III

The type III hand was formerly described as hoof or rosebud shaped (Fig. 45-6). It is the most severe and the least common type. The first web has complex bony or cartilaginous syndactyly, the second and third webs have complex syndactyly, and all four (and often all five) digits have a single, conjoined nail. The thumb nail is small, and, in some cases, distinguishing the thumb from the index finger may be difficult. The syndactyly of the ring and little fingers is simple and

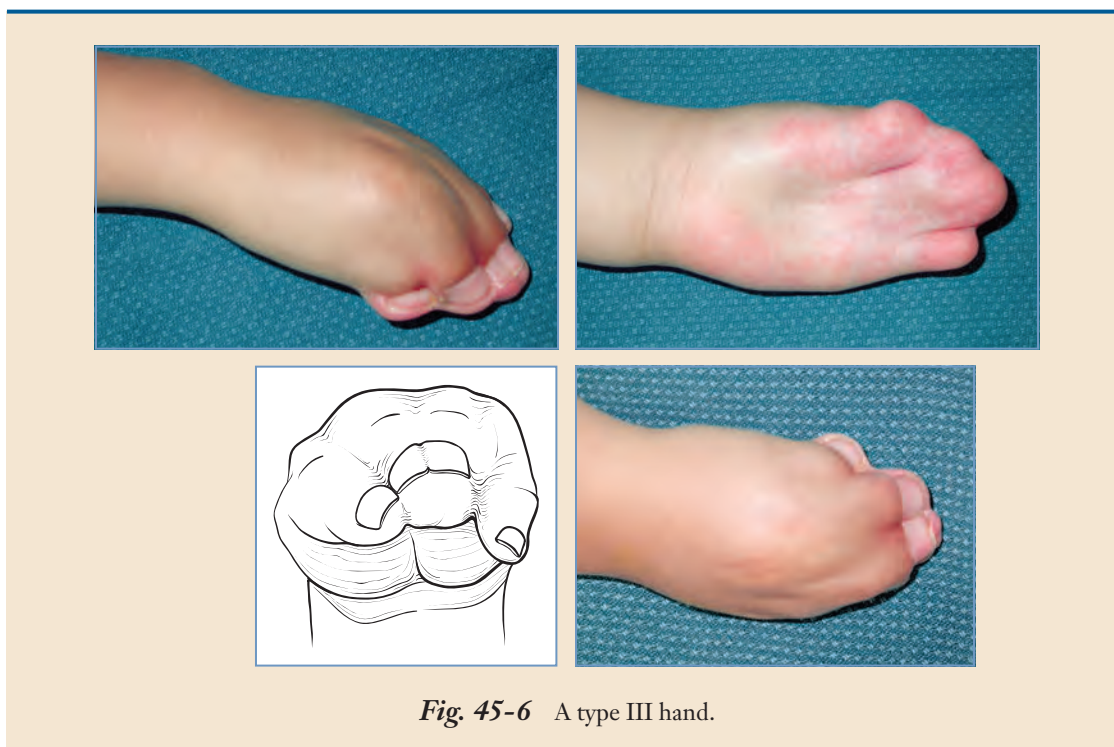


Fig. 45-6 A type III hand.

usually complete. A synostosis of the ring and little finger metacarpals may be present with associated limitation of mobility at the carpometacarpal joint level.

PREOPERATIVE EVALUATION

Evaluating the child in the newborn period and coordinating all surgical consultations and interventions are imperatives. A multidisciplinary team should be involved, including geneticists, pediatricians, anesthesiologists, physical therapists, social workers, and many others.¹¹

Airway problems and feeding difficulties are initial priorities. An upper limb intervention has to be coordinated with complex craniofacial procedures; a craniofacial coordinator usually assumes the clinical lead for planning surgical procedures.

The goals for hand function and the treatments that may be possible are determined. The hands are assessed during the early craniofacial assessment period. Radiographs of the hands are obtained at about 6 months of age to determine the extent of bony anomalies. CT scans with three-dimensional reconstruction may be helpful in type III patients because of the complexity of the anatomy. Routine laboratory tests are performed preoperatively to obtain hemoglobin and electrolyte levels.

TREATMENT OPTIONS

Nonoperative Treatment

Parents should be advised to maintain their child's upper limb passive range-of-joint motion beginning as early as possible. This complements later surgical intervention rather than substitutes for it. Hand therapy with appropriate thumb splints is part of the postoperative treatment phase.

Operative Treatment

Four broad categories of operative correction exist:

1. Digit separation by syndactyly release of all involved web spaces
2. Correction of clinodactyly
3. Release of metacarpal synostosis
4. Revision surgery

Traditionally, the border digits are separated first, particularly the thumb.

SURGICAL PLANNING

Operative Goals for the Hand

The ultimate goal of surgery is a hand that functions and appears as normal as possible. A hand with five digits and adequate web space formation is difficult to achieve. However, this should be the goal to allow optimal use.

Surgery should facilitate growth, and surgical scars should be placed to prevent growth limitation. A goal of surgical planning should be a minimum number of interventions, because these children undergo many procedures throughout childhood. Procedures should be planned and selected to require minimal revision. Surgery to both limbs and to a combination of web spaces can be performed with the patient under the same anesthetic to minimize the number of operations required. A further goal is to complete the hand surgery before patients are 5 to 6 years of age, when possible.

SURGICAL TECHNIQUE

Syndactyly Release

Type I Hand

For patients with type I hands, no treatment of the first web may be required if the web space is of adequate depth and full thumb mobility is possible. Bilateral release of the second and fourth web spaces may be carried out at 6 months of age. The syndactyly is simple but complete in most cases. The commissure is reconstructed using a dorsal flap (Fig. 45-7, A through C).

The dorsal flap needs to be a little longer than in cases of simple syndactyly because of the unusual thickness of the volar skin. Interlocking zigzag incisions are made along the fingers to prevent linear scars and to minimize the potential for scar contracture.^{12,13} Full-thickness skin grafts are taken from the lateral groin or lower abdomen to resurface the resultant defects. Careful selection of donor-site skin prevents the appearance of unwanted hair growth on the hand.

The middle and ring finger syndactyly is usually treated 6 months after release of the second and fourth webs. Particular problems occur during separation of the third web in Apert patients. Only rarely does adequate soft tissue remain over the bone and interphalangeal joints after this separation. Synostosis of the distal interphalangeal joints and fusion of the distal part of the middle phalanges are common. The proximal and middle phalanges are usually symphalangized, and separation of the digits results in a significant amount of bone exposure in the areas of bony fusion over the distal interphalangeal joints.

Some surgeons advocate the use of double-opposing triangular pulp flaps to re-create the nail folds¹⁴ or toe pulp grafts to cover the entire bony defect. This may result in rather pointed fingertips with some nail distortion. Zuker et al¹² stated that the use of pedicled distant flaps was a better option because it has less risk of scar contracture over the distal joint and subsequent deviation with growth. Pedicled groin flaps are individually designed to cover the defect and to



Fig. 45-7 A and B, Operative markings for division of the second and fourth webs. C, Intraoperative view with flaps and grafts inset. D, The third web is divided along its distal two thirds with straight-line skin incisions and the fingers extended to length with K-wires. E, The third web (to be covered) and with a paper template used to aid markings (left). Operative markings for raising a right groin bilobed flap (right). F, A bilobed groin flap is raised. G, The hand inset to the pedicled flaps, with primary closure of the adjacent donor site. H, The long-term result of a groin flap cover over the distal part of the middle finger, with a skin graft placed more proximally.

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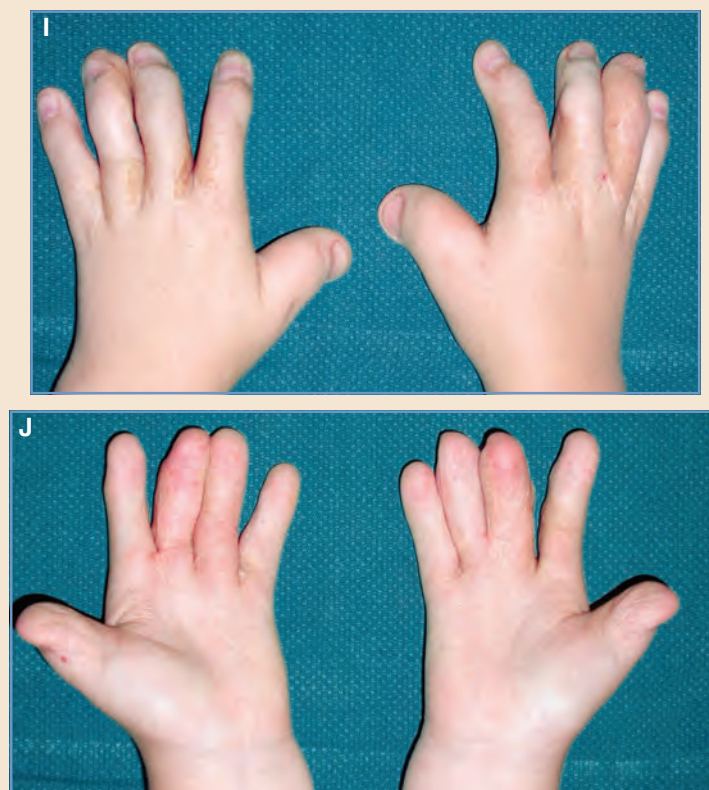


Fig. 45-7, cont'd The long-term outcome in a type I hand. **I**, Dorsal view. **J**, Palmar view.

provide vascularized tissue over the bone, epiphyseal plate, and joint, and appropriately bulky tissue to reconstruct the pulp. The donor site can be closed primarily. In this situation a straight-line syndactyly release is performed in the distal portion of the fingers, with an osteotomy of the fused phalanges. A bilobed groin flap may be designed to cover adjacent finger defects. These areas are divided 10 days later, when the pedicle is divided (Fig. 45-7, *D* through *H*).

The aim of flap coverage is to provide sufficient soft tissue coverage to minimize both scar contracture over the epiphysis and subsequent problems with growth disturbance and deviation. The whole web is not covered with a flap, because this would necessitate a rather bulky, long flap with potential viability problems.

Bilateral pedicle flap surgery is inappropriate, because of the required immobilization and the older age of the child when the third web is treated. Thus some additional surgical procedures are necessary. Six months after the flap is inset, the remaining proximal syndactyly is divided using a dorsal flap technique to reconstruct the commissure, with interlocking zigzag incisions and full-thickness skin grafts to cover exposed areas. The long-term outcome in a type I case is shown in Fig. 45-7, *I* and *J*.

Type II Hand

In type II hands, bilateral release of the first web is the first priority where this is indicated, usually by 6 months of age. An incomplete soft tissue syndactyly can be released using a four-limb Z-plasty technique (Fig. 45-8).

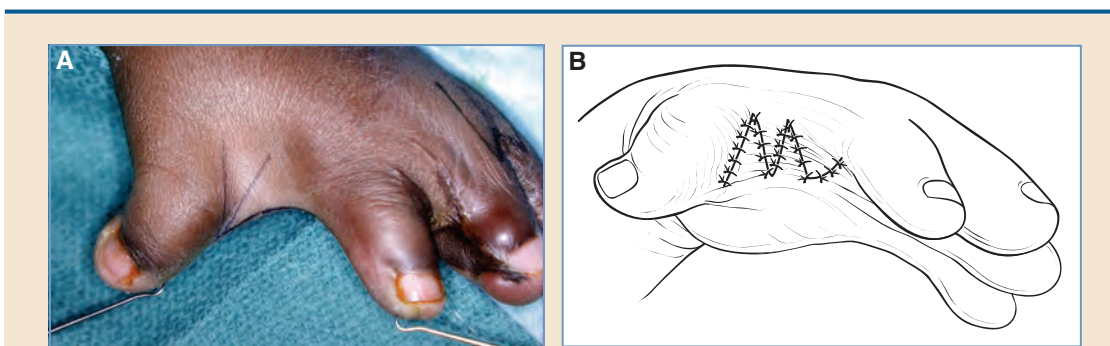


Fig. 45-8 A, Incomplete soft tissue syndactyly of the first web. B, A four-limb Z-plasty to deepen the first web.

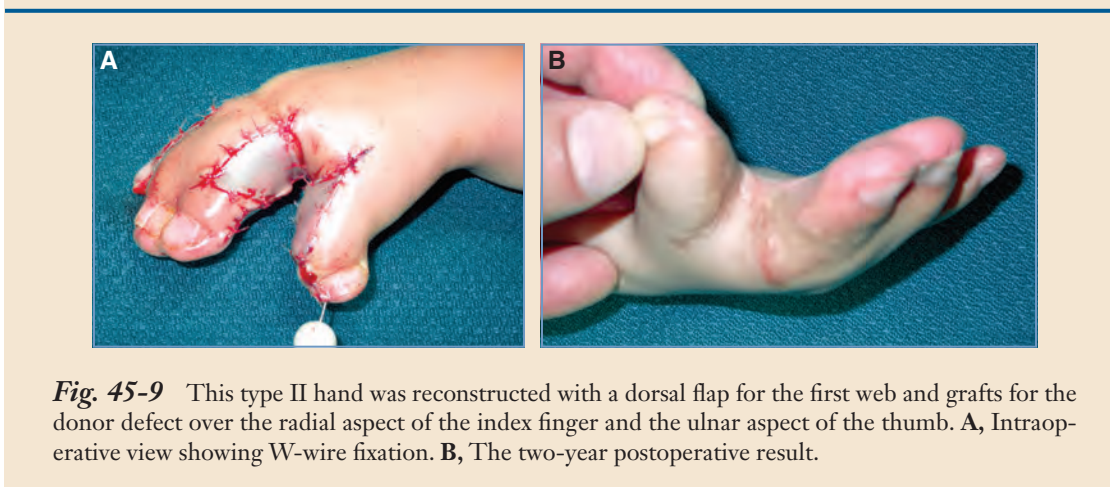


Fig. 45-9 This type II hand was reconstructed with a dorsal flap for the first web and grafts for the donor defect over the radial aspect of the index finger and the ulnar aspect of the thumb. A, Intraoperative view showing W-wire fixation. B, The two-year postoperative result.

The abnormal fascial bands that attach the thumb and index finger are released, along with the investing fascia over the first dorsal interosseous and adductor pollicis muscles. If the syndactyly is more severe, a dorsal first web space flap may be necessary. This can be done by taking a large dorsal flap that leaves a donor defect on the dorsum of the index finger (requiring a skin graft) or by using a dorsal V-Y advancement flap^{9,11} (Fig. 45-9).

Some surgeons have used tissue expansion successfully to provide adequate skin for first web reconstruction.¹⁵ A small expander with an externally placed port is inserted early in life, and reconstruction is performed before the child is 12 months of age. This technique is not widely favored, however. Distally based pedicled radial forearm fasciocutaneous flaps in this situation have been described but are generally reserved for the correction of secondary deformity in adult Apert patients.⁹

Bilateral fourth web release is performed during the same surgical episode as release of the first webs.

Release of the third webs can begin at 12 months of age. Initially, on one side, the distal part of the web is divided, and the deficient soft tissue over the bone and joint is reconstructed with a pedicled groin flap. The flap is divided at 10 days. The proximal web is deepened further at the pedicled flap reconstruction of the opposite side. The final web deepening is performed about 6 months later, when the child is 2 years of age. The second web may be released in the

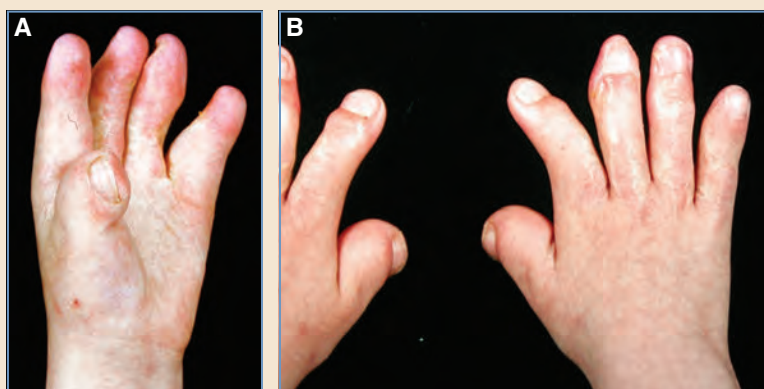


Fig. 45-10 The long-term outcome in a type II hand. **A**, Volar view. **B**, Dorsal view.

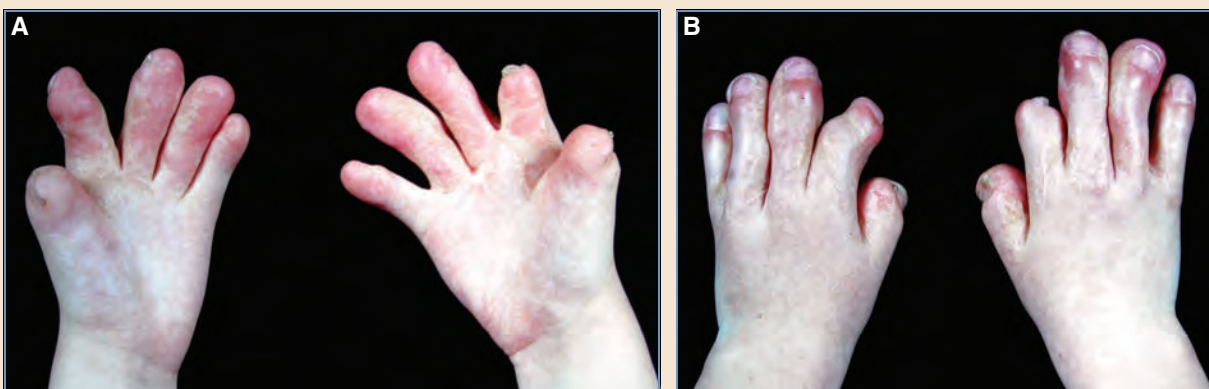


Fig. 45-11 The long-term outcome in a severe type II hand. **A**, Volar view. **B**, Dorsal view.

opposite hand during the flap reconstruction. Long-term results in a type II case are shown in Figs. 45-10 and 45-11.

Type III Hand

Early drainage of paronychia infections may be necessary in a type III hand where pockets of macerated, compressed pulp tissue within the unified nail area have become infected. This may be needed early in the newborn period. Assessment of the bony anatomy can be difficult in these patients, and a preoperative CT scan with three-dimensional reconstruction is helpful.

Some surgeons advocate excision of the whole index ray at the first web reconstruction to ensure a better first web span. However, when this is done, the goal of a more normal hand, with four fingers and a thumb, cannot be achieved. It seems that the initial benefits for hand function are not great, and the long-term sequelae of metacarpal instability are difficult to manage.

Bilateral thumb-index separation is carried out when the patient is 6 months of age at the same time as separation of the ring–little finger web. The first web always has soft tissue deficiency, and a dorsal flap is used to reconstruct it. A proximally based flap from the base of the first to third webs is raised, preserving the dorsal branches of the radial nerve. Skin grafts

are necessary to cover the donor skin deficiency over the first dorsal interosseous muscle. The thumbnail is often small and difficult to delineate from that of the index finger.

Release of the second and third web space on opposite hands may begin at 12 months of age. Release of the third web is performed as described previously, with a combination of pedicled flaps in a distal position, web reconstruction with a dorsal flap, and full-thickness skin grafts to the proximal areas interspaced with interlocking zigzag flaps.

Clinodactyly Release

Thumb radial clinodactyly is usually corrected when a child is 3 to 5 years of age. The thumb is short, broad, and radially inclined because of the presence of a triangular proximal phalanx and a wide terminal phalanx. The deformity becomes more prominent with growth. A longitudinal epiphyseal bracket on the radial aspect of the proximal phalanx, which combines the proximal and distal joint surfaces, is responsible for the triangular shape of the phalanx and the radial deviation of the digit that occurs with growth. With growth and maturity the interphalangeal joint usually becomes fused. Before surgery, hand therapists assess the child's function, and, with the surgeon, decide whether the child's function is likely to be improved with an osteotomy. Many of these children may not need surgery for this functional (and aesthetic) issue.

An opening wedge osteotomy to improve length and correct angulation is the operation of choice for thumb clinodactyly.¹⁶ The surgeon approaches the area through a radial Z-plasty incision situated as distal as possible along the midaxial line. This allows some soft tissue elongation. Care is taken to preserve dorsal sensory nerves and veins. The interphalangeal joint is located by passive motion, and the attachment of the abductor pollicis brevis and radial collateral ligaments needs to be lifted to access the phalanx. The bracketed epiphysis is usually situated more to the ulnar side of the metacarpophalangeal joint and is difficult to access. Osteotomies on these small bones are challenging. The surgeon performs a transverse and distally positioned osteotomy, leaving the ulnar-side periosteum intact and removing a wedge of bone. This is rotated 180 degrees and inset into the osteotomy site (Fig. 45-12).

A bone graft is usually required, and different sources are available. Good-quality cortico-cancellous bone can be obtained from the iliac crest, but sufficient bone may be found at the site of division of the fourth and fifth metacarpal synostosis and can be obtained during the same surgical procedure.

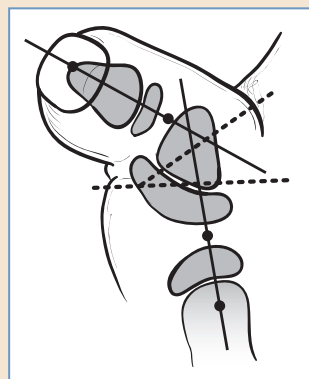


Fig. 45-12 Osteotomy planning for correction of thumb radial clinodactyly.

The surgeon immobilizes the area with paired longitudinal K-wires. Soft tissue elongation is achieved through Z-plasty alone, although the addition of skin grafts may be necessary. The K-wires are removed after bony union has been confirmed, but further support with a splint is continued for 2 to 3 months.

Both the thumb and index finger may be effectively straightened with this type of procedure, which, in appropriate cases, can improve pulp-to-pulp pinch and grip strength. The index finger may also be radially inclined because of the presence of abnormal skeletal components. A radial longitudinal epiphyseal bracket of the proximal phalanx, which articulates with the distal phalanx, may be present, because of symphalangism of the proximal and middle phalanges. After division of the second web, and with growth, the deformity becomes visible. If a split epiphysis is present, the radial part does not contribute to the longitudinal growth of the phalanx. The middle phalangeal bone may be abnormal and triangular and a main factor contributing to abnormal growth of the digit. The use of an osteotomy and distraction of the proximal phalanx to correct thumb clinodactyly has been described as a successful technique in a small number of patients.¹⁷

Metacarpal Synostosis

Synostosis of the fourth and fifth metacarpals causes a flatter configuration of the hand, restricting movement of the fifth ray. Synostosis of this kind is present in two thirds of Apert patients.⁸ An osteotomy and excision of bone joining the two metacarpals can improve grip strength and span of the hand.

If division of synostosis is performed at the thumb clinodactyly correction, bone graft material may be available for the thumb opening wedge osteotomy. However, early correction of synostosis has not been successful; the procedure should not be performed in patients much younger than 5 years of age to prevent re-formation of the synostosis. Insertion of an intervening buffer material is required to prevent re-formation. Various synthetic and autologous materials have been used, and Upton⁹ advised using deepithelialized skin. Release of the dorsal capsule of the carpometacarpal joint and/or the ligament attachments provides mobility and rotation capabilities at this joint.

POSTOPERATIVE CARE

A secure dressing, usually including a plaster cast, is essential after complex surgery on the hand, particularly in children younger than 2 years of age. The dressing should extend above the elbow, with the elbow in a partly flexed position, if it is to remain in place for at least 10 days for skin graft protection.

When pedicled flaps are used, the hand and arm have to be immobilized to the chest wall for approximately 10 days. This is achieved ideally with a well-fitting and well-padded soft bandage rather than a plaster cast. Plaster casts in this situation can lead to pressure sores and skin irritation or laceration. Children appear to tolerate this remarkably well.

REVISION SURGERY

Although the aim is to allow these children to have four digits and a thumb on each hand by school age, further surgery is often required, especially for type III hands. This may be needed to improve appearance and function. The appearance of the hands is important. During the teenage years concerns about visible scars, hyperpigmented skin grafts, and broad nail folds may arise. Redeepening of interdigital web spaces, reduction of soft tissue bulk, and excision of hyperpig-

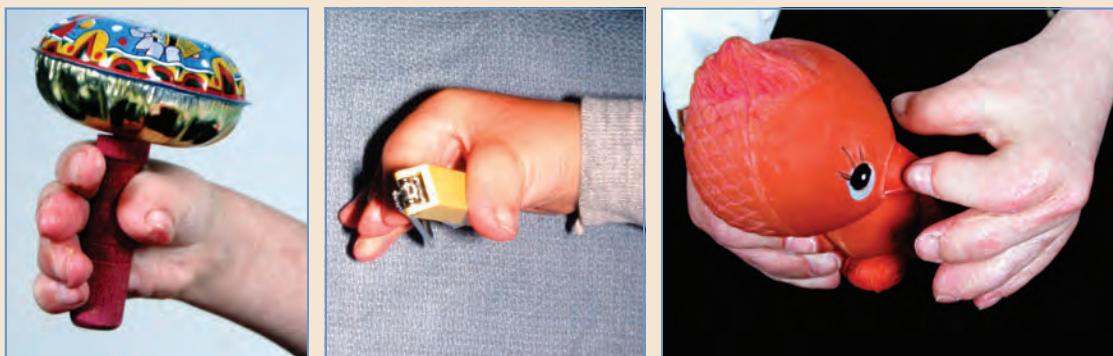


Fig. 45-13 Final outcome showing a range of ability to grasp small and large objects with thumb–index finger precision.

mented and hair-bearing skin grafts may be required. Z-plasty for scar contracture may be necessary. The appearance of the nail folds can be improved by strip excision of the nail folds and eponychial fold reconstruction. For functional improvement, an osteotomy to correct angulation or rotation may be necessary. Fearon¹⁸ advocated midphalangeal osteotomies of the four digits to improve thumb-to-finger pinch and grasp.

TREATMENT OUTCOMES AND COMPLICATIONS

Minor areas of skin graft loss and tiny areas of marginal flap necrosis can occur. Major complications are rare. With expert anesthesia care, respiratory problems are unusual. Major infection complications and partial and complete digit loss are rare, although families should always be warned about these possibilities. In general, patients and families are pleased with a four-fingered hand that functions remarkably well despite the bony and cartilaginous limitations. It should have an adequate first web space, sufficient soft tissue to prevent wound breakdown, and the ability to pinch and grasp (Fig. 45-13).

ALTERNATIVE SURGICAL TREATMENT MODALITIES

Apert¹ attributed some observed delay in these children's development to their musculoskeletal anomalies. This observation may have prompted the need for early intervention, combining the correction of both the craniofacial and upper limb anomalies. Upton⁹ described three essential concerns in the upper limb: (1) skeletal unions, (2) failure of complete joint segmentation, and (3) incomplete separation of the digital rays. Traditionally, digit separation gives priority to the separation of the border digits, particularly the thumb, as described previously.¹⁶

Chang et al¹⁷ reported a simplified traditional approach. In their series, syndactyly release was performed using local flaps and full-thickness skin grafts taken from the groin in all cases, without the need for distant flaps. They also carried out a thumb osteotomy in conjunction with syndactyly, using the ulna as a bone graft donor site. The digits were separated and the thumb clinodactyly corrected by 2 years 5 months of age. However, the authors did not attempt to create five-digit hands in the type III cases in this series.

Fearon¹⁸ adopted a rather different approach for separating all five digits in each of the four limbs in two operations. This approach produced a different philosophy of timing of intervention and technique. The first phase of Fearon's technique uses a straight-line skin incision release of each syndactyly. His technique does not give preference to the border digits, allowing separation of nonadjacent webs and preventing difficulties with vascularity of the digits. It does not attempt to cover exposed bone or joint in the distal half of the interdigital space with additional grafts or flaps. Instead, these areas are left to heal by secondary intention. However, the proximal web is reconstructed using a combination of volar and dorsal flaps and full-thickness skin grafts taken from the lower abdomen.

Fearon's second phase of treatment evolved as a result of functional difficulties with daily activities such as dressing. During the second phase the surgeon performs osteotomies of the phalanges midway between the metacarpophalangeal joint and the distal interphalangeal joint of each finger. An opening wedge is performed in each case, and the digits are angulated to allow pulp-to-pulp pinch with the thumb. Thumb clinodactyly correction with bone grafting is performed at the same stage.

Matsumoto et al¹⁹ described the correction of thumb angulation and length in two Apert cases using an external ball-jointed distraction device. An osteotomy of the proximal phalanx was initially performed. After a 1-day waiting period, early distraction followed by 1 mm of daily lengthening with alteration of the external angle achieved satisfactory alignment without the need for bone graft or interference with the epiphyses. Premature bony fusion at the osteotomy site occurred in the first of their Apert cases, prompting a change in protocol.

Fragale et al²⁰ studied in vitro osteoblast cultures taken from skull bone samples of clinical craniosynostosis cases, including one Apert case. The craniosynostotic osteoblasts showed a lower proliferation rate than that of controls. They suggested that unorthodox *FGFR2* activation may play an important part in this.

Traditionally, division of toe syndactyly is not routinely performed but may provide a significant psychological benefit rather than a functional one. Fearon¹⁸ advocated it for this reason.

In the last 5 years our group and others have begun offering simultaneous release of all web spaces to highly selected patients.^{21,22} We do not offer this approach routinely, but only after certain criteria have been met.

Conceivably, all patients with multiple, adjacent, web space syndactylies could undergo release in a single surgical stage, including those with Apert syndrome, provided the skin coverage and vascular supply to each ray are sufficient. We have used this approach in 14 hands thus far, and we have been impressed with the level of satisfaction the families have expressed. We think that, for certain families, this simultaneous release approach is a valuable alternative to a conventional, staged approach. The main benefit of a single-stage approach lies in the potential to combine multiple stages into fewer (ideally one) operations. This approach may be especially relevant for families of Apert syndrome children. Because of their combined craniofacial and limb anomalies, most of these children undergo more than 20 procedures, each requiring a general anesthesia, by the time they are skeletally mature. Not only does this large number of surgeries present a logistical challenge to families and surgeons, particularly for those traveling from far away, it also raises an emerging concern that repeated general anesthetics in young children may impair neurologic development. The psychological impact of enduring multiple sequential operations and rehabilitation protocols in complex syndromes should not be discounted.

The first step in determining which families would benefit from simultaneous release involves the input of the hand therapist. He or she must critically evaluate the ability of the family

to endure what may be a more arduous course of postoperative rehabilitation, stretching, splinting than may otherwise be necessary with a series of smaller operations. This decision is largely based on judgment and experience.

If the therapist and surgeon concur that the family can tolerate the rehabilitation, then arterial phase CT angiography is performed. We use a special protocol to reduce radiation exposure to very small levels roughly equivalent to those of a conventional chest radiograph. Sedation may be needed for the study, which can often be combined with a craniofacial CT as needed. We review the CT angiogram with our radiologist to determine whether each ray has at least one robust-appearing artery and simultaneously formulate an operative plan. The family should be informed that, although no cases of digital loss have been reported so far, such a loss may occur if vascularity is inadequate.

If we think that a child is a good candidate for simultaneous release, then we plan to release all webs during one anesthetic event. We use a two-team approach to minimize operating time. Cases of 10-digit syndactyly require more than 6 hours of total operating room time. We use a hand table on one side and a standard operating table on the other. Bilateral upper arm pneumatic tourniquets are used, and intravenous access is obtained with lower extremity lines. The entire lower abdomen is prepared and draped for full-thickness skin graft harvesting, which will leave a transverse scar from hip to hip in cases with all webs involved.

The design of the flaps incorporates dorsal quadrilateral flaps and straight-line releases, as with a conventional, staged release. The main difference is that comparatively more skin graft area may be needed if all webs are to be released. However, we have not found this to be a significant functional or aesthetic issue so far.

We generally release the border digits first, especially in type III cases. However, some surgeons prefer to release the first and third webs to mirror one of the more popular conventional approaches. We then sequentially release each web, examining the neurovascular bundles in each case. A CT angiogram accurately predicts the presence of arteries, but the test is not sensitive enough to detect very small arteries in some cases. In all cases we have treated thus far, every digit has had at least one major artery, and most have had two. Once the release is completed, we obtain templates of the defects, transfer them to the abdomen, and harvest full-thickness skin grafts. Usually, no undermining is needed to close this donor site, but cases that require a large amount of graft might require undermining of the abdominal skin to facilitate closure. We secure the grafts with 6-0 polylactic-co-glycolic acid (Vicryl Rapide) sutures and close the donor site with layered 4-0 and 5-0 polycaprolactone (Monocryl) sutures.

We used to use hard plaster dressings; however, in the last 5 years we have transitioned to a simpler soft “boxing glove” dressing. This type of dressing is commonly used in Australia and the United Kingdom for syndactyly and other congenital hand surgery but is not often used in North America. We place antibiotic ointment and a nonstick gauze dressing (Bactigras or an equivalent) in the webs to cover the grafts, followed by soft roll gauze (or an equivalent soft absorbent gauze) in the webs. The placement is easier when the fingers are held apart by temporary stay sutures (such as 4-0 silk sutures). We wrap the hand up to the wrist in the same soft roll gauze and clean the upper extremity to remove detergent and any other substance that can interfere with taping. Then, we wrap the hand with sticky tape. (We prefer Hypafix, because it is very sticky yet pliable.) Tape is applied from the level of the hand to nearly the axilla to ensure it remains secure for one week. In the clinic we generally reapply a new dressing 1 week postoperatively. Typically, the healing is adequate by 2 weeks to allow unrestricted motion. If healing is not sufficient, then a lighter dressing, applied in the clinic, may be needed for another week.

Outcomes

We have found that patients are satisfied and function is adequate (Fig. 45-14). Whether the functional and aesthetic outcomes of this approach are truly equivalent to those of conventional, staged surgery is unknown. As documented in our report,¹² we noted one episode of cellulitis requiring IV antibiotics, and one family had difficulty complying with the therapy regimen. One patient required a secondary release for a severe flexion contracture that could not be completely corrected at the initial operation. Overall, our data show a high level of satisfaction with this approach, although we have yet to benchmark this satisfaction level with that of a conventional approach. Nevertheless, the outcomes appear to be overall equivalent to those of a conventional, staged release.

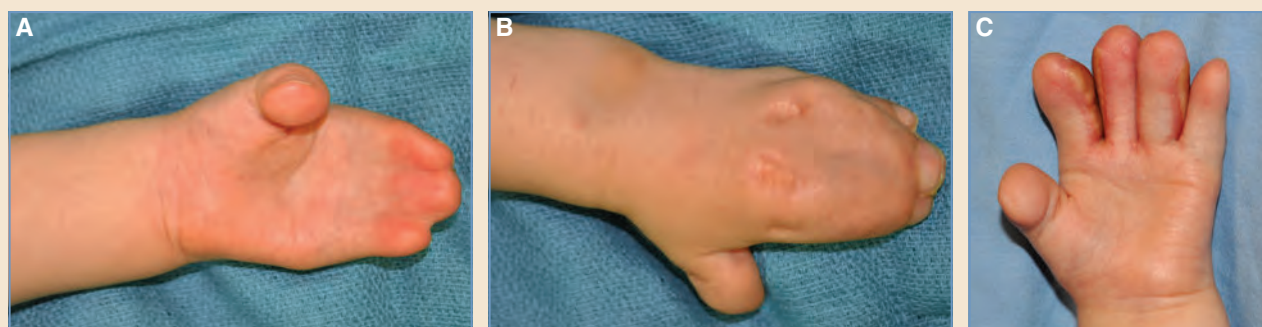


Fig. 45-14 A and B, Preoperative volar and dorsal images of a type II hand. C, A postoperative image.

KEY POINTS

- Apert syndrome is one of the craniosynostosis syndromes and is associated with symmetrical and severe bilateral syndactylies of the hands and feet.
- The hands in Apert children are strikingly symmetrical. The deformity is complex and has the following characteristics:
 - Complex syndactyly of the index, middle, and ring fingers
 - Simple syndactyly of the ring–little finger web
 - Symphalangism of the index, middle, and ring fingers
 - Short thumb with radial clinodactyly
- Surgical priorities include the following:
 - Digit separation by syndactyly release of all involved web spaces
 - Correction of clinodactyly
 - Release of metacarpal synostosis
 - Revision surgery

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46

Polydactyly

John A. Girotto • Rajiv Sood

DEFINING THE PROBLEM

Polydactyly is the presence of more than five digits on one extremity. This encompasses many heterogeneous abnormalities, including partial and complete duplication of parts, occasionally with concomitant syndactyly. The word *polydactyly* is often used to describe supernumerary digits, whereas *duplication* usually refers to thumb anomalies. The appearance is easily noticed and difficult to conceal, leading to an immediate cosmetic concern for parents and an eventual cosmetic concern for patients. A rudimentary digit may interfere with the function of the primary digit, depending on the level of duplication, destabilizing the joint by separating its radial and ulnar attachments or by stiffening the joint irreparably. Many of these secondary abnormalities are more thoroughly discussed in other chapters.

In the United States, ulnar polydactyly is the most common congenital hand anomaly, with a prevalence of 1.5 to 1.7 per 1000 live births.¹ It is associated with many syndromes (Table 46-1) but often occurs sporadically.¹⁻⁴ Ulnar polydactyly is twice as common in males than females⁵ and 10 times more prevalent in blacks than in whites.^{6,7} Other polydactylies are much less common in general with a prevalence of 5.4 to 5.7 per 100,000 births.²

Several systems of categorization have been used in the past, including such nonspecific names as *preaxial* and *postaxial polydactyly*. Because of the surgical implications, many groups have suggested systems of categorization based on the bony anatomy. In an effort to form uniform terminology, the International Federation of Societies for Surgery of the Hand has adopted the modern Classification of Duplication (III)⁸ encompassing radial, central, and ulnar duplications (Box 46-1). Radial (or thumb) duplications are further classified most commonly by Wassel types I through VII (Fig. 46-1). This classification is based on the bony level of duplication. An incomplete bony separation of the distal phalanx constitutes type I, whereas a complete separation of the distal phalanx constitutes type II. Similarly, the proximal phalanx duplications are described

Table 46-1 Syndromes Associated With Polydactyly

Syndrome	Hand Findings	Systemic Findings
Ulnar Polydactyly		
Trisomy 13	Polydactyly	Microcephaly and mental/motor challenges Meningomyelocele Cleft palate Eye defects Urogenital defects Heart defects
Bardet-Biedl	Polydactyly, syndactyly	Heart defects Obesity Urogenital disease Mental/growth delay
Smith-Lemli-Opitz	Polydactyly, foot syndactyly	Microcephaly and mental challenges Malformation of the heart, urogenital tract, and gastrointestinal tract
Radial Polydactyly		
Fanconi anemia	Radial hand differences	Pancytopenia Short stature Eye problems Hearing problems Malformation of the heart, lungs, kidneys, and digestive tract
	Townes-Brocks	Imperforate anus Hearing problems and ear-shape malformations Malformations of the heart, kidneys, and genital tract
Holt-Oram	Thumb hypoplasia	Cardiac abnormalities

Box 46-1 Classification of Duplication of the International Federation of Societies for Surgery of the Hand

Radial (thumb)

Wassel types I-VII

Central

Often associated with syndactyly (complex)

Ulnar (fifth digit)

Types A, B

as types III and IV. Wassel type VII deformity, or triphalangism, is a separate entity and refers to an extra phalanx (rather than an extra digit). This phalanx can be rectangular or wedge shaped, and fully developed or hypoplastic, with implications about size, curvature, and related functional problems. Triphalangism with an additional duplicated thumb is most often seen in Wassel type IV (Fig. 46-2, *A*) and Wassel type VII duplications (Fig. 46-2, *B*). In 2008 the Rotterdam group⁹ expanded the thumb duplication classification to include triphalangism.

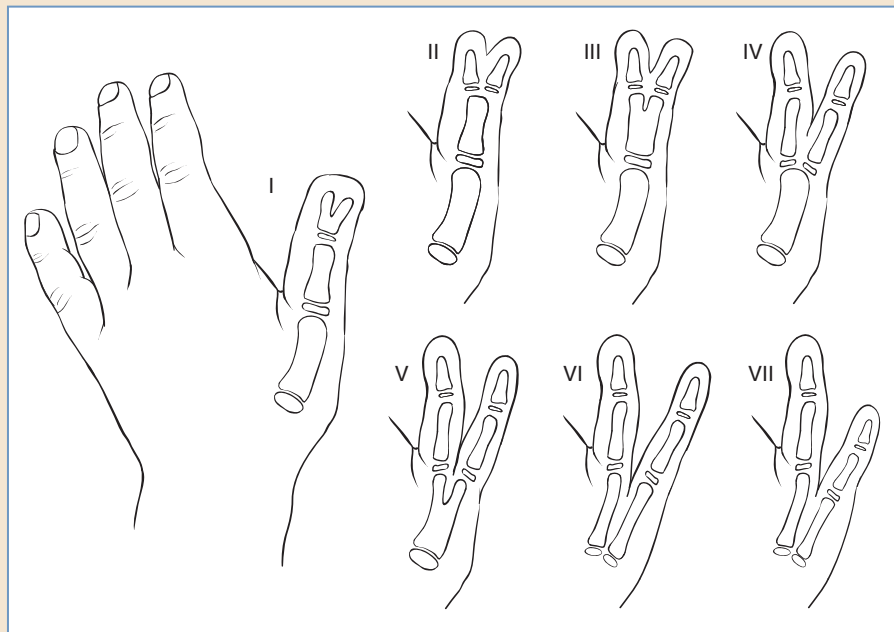


Fig. 46-1 The Wassel classification of thumb polydactyly.

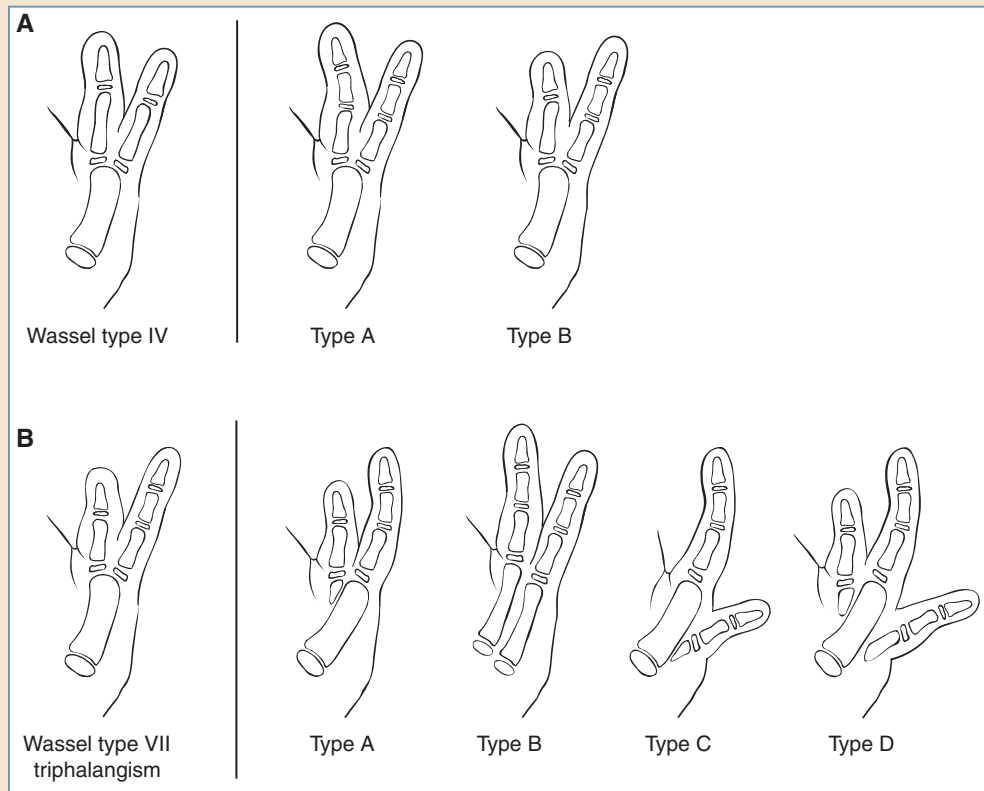


Fig. 46-2 Triphalangism with a duplicated thumb. **A**, Wassel type IV classification, types A and B. **B**, Wassel type VII classification, types A, B, C, and D.

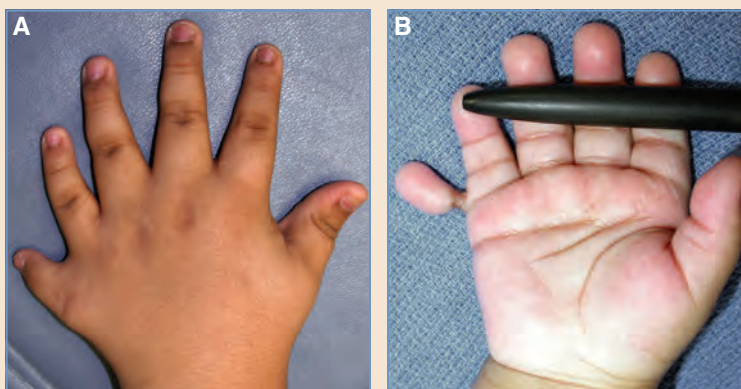


Fig. 46-3 Ulnar polydactyly. **A**, Type A involves a fully developed digit. **B**, Type B involves a rudimentary appendage.

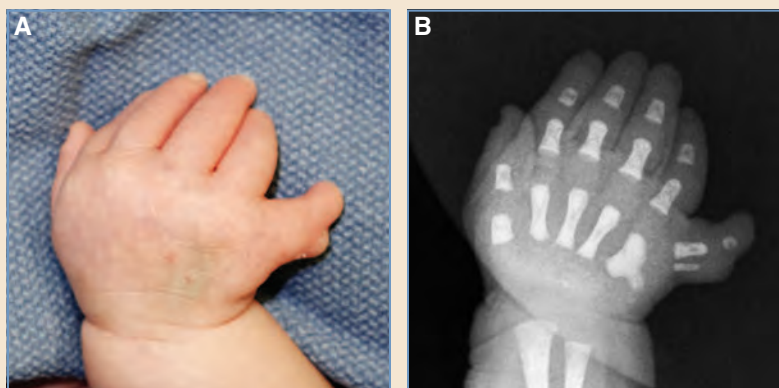


Fig. 46-4 Ulnar polydactyly. **A**, This clinical presentation is not within the classification. **B**, A radiograph of the same patient.

As many pediatric hand surgeons have observed, and as Ezaki¹⁰ has described, thumb polydactyly does not mean that the thumb is really duplicated; it is more correct to describe it as split thumbs at certain levels (that is, Wassel types I through VI). Each one is smaller than the corresponding digit on the unaffected side. In some cases, creating one normal-sized digit by linking the two duplicated thumbs together might be imagined. The structural development problem is not only size related but affects all tissues. Thus the tendons, joints, ligaments, and other tissues distal to the duplication are not normal. Reduced range of motion and joint instability can be anticipated, and patients should be followed for these throughout the growth period.

Ulnar, or fifth finger, duplications are either a fully developed digit (type A) or a rudimentary appendage (type B) (Fig. 46-3). A classification system comprising three types has been described, in which type I correlates with a rudimentary appendage, type II includes duplication of the metacarpophalangeal joint, and type III includes duplication of the entire ray.⁶ Given the variability of expression, some presentations are not represented in standard classification schemes (Fig. 46-4).

Central duplications are rare and often involve syndactyly (failure of differentiation), overlapping their classification. Syndactyly is more thoroughly discussed in Chapter 44, and the classification is discussed in Chapter 41.

Epidemiology

The most significant advances concerning polydactyly involve factors that influence in utero development of fetal limbs. However, the embryologic causes of duplication have not been defined. Despite various systems of classification, treatment has focused on removal of one of the duplicated (rudimentary) parts. More recently, treatment has been recognized as reconstruction, not simply removal, of the affected digit. This includes concomitant joint and tendon rebalancing with removal of the duplicate digit.¹¹

Genetics

Limb development occurs during weeks 5 through 8 of gestation, with limb bud formation and the emergence of the polarizing region of the apical ectodermal ridge.¹² Polydactyly results from defective patterning of the anteroposterior axis of the developing limb. Several *HOX* genes encode critical transcription factors related to the development of synpolydactyly.¹³ Specifically, mutation of *HOXD13* is associated with limb synpolydactyly.^{14,15} The dHAND transcription factor plays a critical role in the anteroposterior orientation of the polarizing region of the apical ectodermal ridge.¹⁶⁻¹⁸ The *sonic hedgehog* (SHH) signal from the polarizing region of the limb bud may appear in an ectopic anterior location, giving rise to radial polydactylous duplications.^{12,19} Mirror anomalies have been experimentally created by moving the SHH signal to an anterior border location.^{12,18}

Transmission of polydactyly varies. A recent meta-analysis of the genetics literature identified 310 different disorders that include polydactyly at presentation and involve mutations in more than 90 separate genes.²⁰ All patterns of inheritance were represented (autosomal dominant, recessive, X-linked, and multimodal), but more than 109 syndromes had an unknown mode of inheritance.

A triphalangeal thumb (TPT) and its related polydactylous forms (that is, Wassel type IV, A and B, and type VII, A through D) seem to be inherited as an autosomal dominant trait. A number of syndromes associated with TPT are possible. A careful genetic evaluation should be considered before surgical intervention. In families with inherited but isolated radial polydactyly, a link to chromosome 7q36 has been identified that affects protein SHH regulation.

Alternatively, central polydactyly has a definite autosomal dominant inheritance pattern but with wide variation in its expression. This may include complex syndactyly, which in this situation is often referred to as *polysyndactyly* or *hidden polydactyly*.

Ulnar polydactyly has a definite inheritance pattern, but it is more complicated than that of central polydactyly. When ulnar polydactyly is an isolated malformation, the inheritance pattern is autosomal dominant. This is commonly seen in blacks with the type B form. However, penetrance is variable, making tracking a direct lineage difficult. Alternatively, when ulnar polydactyly is associated with other anomalies or, as seen in whites with the type A form, the inheritance pattern is autosomal recessive, a careful genetic evaluation should be considered.

PERTINENT NORMAL AND ABNORMAL ANATOMY

Functional Implications

Duplicated digits often have anomalous connections of collateral ligaments, leading to joint instability. This is corrected by surgically reattaching the affected collateral ligament to stabilize the joint. For thumb and small finger duplications, the abductor inserts on the lateralmost digital remnant, thus pulling it into further abduction and destabilizing the joint. In turn, this position influences the developing shape of the distal aspect of the first normal bone on which the duplication rests. The abductor insertion and the joint shape must be surgically corrected to centralize and stabilize the reconstructed digit. Duplications within the joint can limit motion, leading

to a stiff joint that persists even after reconstruction. Ultimately, the bulk and orientation of the duplicated parts may block thumb-finger opposition. Surgical intervention should begin early with duplications that clearly interfere with thumb opposition.⁵

Cosmetic Implications

Our hands are rarely concealed with clothing and are often the first thing seen by another person. The cascade of fingers is a pattern quickly recognized by parents and strangers. The cosmetic implications of extra or duplicated digits are undeniable. Infants quickly learn to function with whatever parts of the extremity they have present, but when they are with other children they may be scrutinized and ridiculed. Treatment, which may involve staged operations, is often planned to conclude by the time the child begins school.^{5,21}

PREOPERATIVE ASSESSMENT

History and Physical Examination

Because polydactyly has been associated with multiple genetic syndromes, a careful and thorough evaluation is warranted in even the simplest cases.^{3,4} A history of hand abnormalities in siblings or the parents may shed light on a familial predisposition. If an associated syndrome is suspected, genetic testing and pediatric consultation are warranted.

Evaluation and Workup

Careful observation of the motion and function of the affected hand/digits should help to determine opposability and supple motion of an affected joint. The development of the affected digit in relation to adjacent digits is noted, including excess or hypoplasia of these structures and of the other hand. The condition and size of the nail and the quantity of soft tissue available are noted for reconstruction. In a Wassel type IV duplication, the metacarpal is often broad (broader than the one on the normal side), and the proximal phalanx that will be preserved is typically narrow (narrower than the one on the normal side). This relationship should be pointed out to the family, because it will be a noticeable residual asymmetry (relative to the normal side) after surgical intervention. Joint alignment, lateral stability, and bony angulation need to be assessed preoperatively and addressed at the initial operation. Last, the tendinous insertions must be noted on the anomalous parts to plan balancing and reinsertion on the reconstructed digit that remains.

Laboratory and Imaging Diagnostics

At a minimum, AP and lateral radiographs are obtained despite the child's skeletal immaturity. These views demonstrate baseline anatomy for classification and may be a critical guide to eventual reconstruction. Although discrete bones may not be identifiable, widened gaps between visible bones can indicate the position of aberrant bone in the future.²² This is true for all radial and central duplications and ulnar type A duplications.

Isolated ulnar polydactyly, however, is often more benign, and radiographic evaluation in children with a type B presentation may not be required.

NONOPERATIVE AND OPERATIVE TREATMENT OPTIONS

Indications

Reasons not to intervene surgically in the presence of polydactyly are rare. In most cases the supernumerary digit or the duplicated digit poses a cosmetic problem (as in type B ulnar polydac-

tyly), a functional problem (as in types IV through VII radial polydactyly), or both and requires surgical extirpation with reconstruction. Type B ulnar polydactylies are often treated by ligation in the newborn nursery. Reviewing this less invasive approach, Mills et al²³ examined 231 hands with at least 2 years of follow-up and found only 7% underwent surgical revision after ligation in infancy.³ Although effective in most cases, this practice can be complicated by infection, vigorous hemorrhage, and a commonly reported ulnar nipplelike remnant with a painful or hypersensitive neuroma.²⁴

Contraindications

The only contraindications to surgical reconstruction are concomitant congenital abnormalities that preclude an anesthetic induction.

PREOPERATIVE PLANNING

Operative Treatment

Ulnar supernumerary digits (type B) require only excision with control of the digital artery and careful division of the digital nerve below the surface of the skin. They do not involve aberrant tendinous connections and rarely involve the bone or joints. The skin is closed primarily over the ligated neurovascular bundle to prevent a painful, superficial neuroma.²⁴

Other duplications—on either the radial or the ulnar side—may necessitate removing the duplicated segments or combining the duplicated parts to achieve the best functional and aesthetic result. This requires restoring joint balance and stability by reattaching collateral ligaments, reefing the joint capsule, or a bony osteotomy.

The guiding principle is restoration of the axial balance of the digit laterally, volarly, and dorsally. This usually involves addressing all of the tissues—skin, tendon, ligament, and bone/cartilage.

Timing

The timing of surgery has been somewhat arbitrary. Most authors recommend surgical reconstruction before the first birthday. In ulnar polydactyly, simple type B cases can be performed in the nursery or during the newborn period. The more complex type A cases with bony attachments can be treated at about 6 to 9 months of age. Similarly, most thumb duplications can be treated when the patient is just younger than 1 year of age. At this age, the structures are large enough to identify and adjust, the anesthetic risks are low, and fine hand prehensile function has not yet developed. Complications such as joint instability, axial deformity, and infections occur more frequently in older children.²⁵ Rarely, if unsure of which digit to remove, the surgeon should delay surgery until function and usage can be assessed. This may take additional time to clarify.

Patient Positioning and Marking

Similar to other pediatric hand cases, preparation includes the use of an arm board, a tourniquet, loupe magnification, and general anesthesia. Unlike adult patients, who usually care for themselves, very young children receive total care from a caregiver; thus surgeons can correct both hands simultaneously, if necessary. Zigzag incisions are used to prevent linear scars, which cause contracture and growth disturbance during development. Border racquet-handle incisions work well when possible. The thumb is often approached from the radial side to preserve the metacarpophalangeal ulnar collateral ligament. Most thumb duplications are asymmetrical, with the radial digit being smaller and often nonfunctional. In this case, the radial digit is clearly the one

to remove. Approaching this digit from the radial side allows access to the structures that require exposure and reconstruction (the radial collateral ligament, the extensor and flexor tendons, and the bone and joint), preserving the ulnar structures. Careful marking should maximize preservation of hearty soft tissue coverage. Excess tissue can be trimmed during closure.

SURGICAL TECHNIQUE

Ulnar Polydactyly

Ulnar supernumerary digits with only a rudimentary appendage (type B) should be excised through a racquet-handle incision around the base of the supernumerary digit. The neurovascular structures should not be divided prematurely. The skin is incised, and the artery and nerve immediately below are identified. With continued dissection, the branch point of the fifth ulnar digital nerve and the supernumerary bundle can be seen. Bipolar cautery is used to divide the artery and provide hemostasis. The nerve is sharply divided below the skin level, and great care is taken to not pull up an attached segment of the normal digital nerve during the division. This catastrophic error leads to ligation or division of the fifth finger's ulnar digital nerve, resulting in anesthesia of that side of the digit. After division, the supernumerary digit is removed, allowing primary closure of the skin²⁴ (see Fig. 46-4).

Treating a type A completely duplicated ulnar digit is more complex than treating a type B pedunculated stalk.²⁶ Similar to thumb duplications, these complete fingers may have muscular insertions from the hypothenar muscles and collateral polydactylous ligaments that must be addressed with reinsertion. Unlike the thumb, the fifth finger is not often used in opposition or stressed laterally, gripping objects. The remaining finger must be strong in flexion, with axial stability. If the duplication extends from the level of the proximal phalanx, the abductor may require preservation and reinsertion (Fig. 46-5). If the duplicate is more proximal, aberrant insertions of the flexor brevis or opponens may require subperiosteal elevation and connection to the remaining digit.

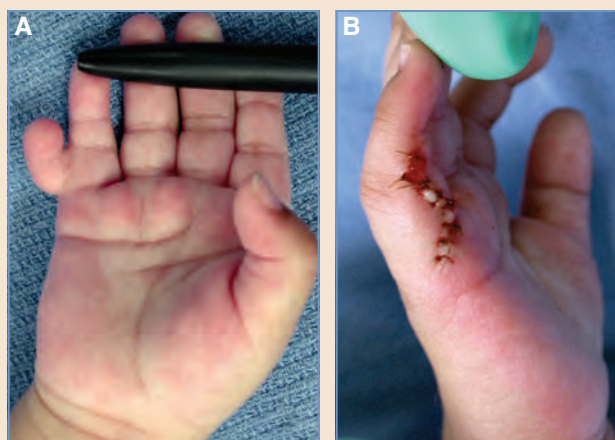


Fig. 46-5 Type A ulnar polydactyly at the level of the proximal phalanx. **A**, The preoperative appearance. **B**, A zigzag lateral incision to facilitate exposure of involved structures.

Central Polydactyly

Duplicates that extend from the central digits are quite variable, and it is difficult to generalize regarding their specific treatments.²⁶ An element of syndactyly obscures the anatomy of the duplicate part and identification of the dominant digit (Fig. 46-6). Similar to other syndactylies, a paucity of skin is present despite an overall excess of tissue. Incisions should be carefully designed to prevent straight-line scars that tether developing digits, and all usable skin should be saved for closure. Surgeons must evaluate duplicate tendinous units and insertions. As with all polydactyly treatment, basic tenets should be followed, specifically:

- Preservation of skin
- Alignment of bone and articular surfaces
- Rebalancing of tendons and muscles

When duplicated central digits are removed, as with other ray amputations, the space that remains must be narrowed by approximating the intermetacarpal ligaments.

Radial Polydactyly

The aim of surgical reconstruction is to obtain a stable, yet mobile thumb of adequate size and shape. Stability and size relate to strength, both for grip and pinch. Thumb mobility is largely dependent on the integrity of the carpometacarpal joint. This is often normal in Wassel types I to IV. The range of normal metacarpophalangeal joint motion is variable in all people, specifically in the degree of flexion. Loss of interphalangeal joint flexion impairs tip pinch to some degree, but children compensate effectively without appreciating any significant loss of function.

Wassel Types I and II

In most cases of Wassel type I and type II polydactyly, the ulnar digital component is larger and more functional and should therefore be preserved. Care is taken to ensure adequate tendon function and to restore the radial collateral ligament as the interphalangeal joint is repositioned. In general, it is easier to repair the collateral ligament after the joint has been aligned and maintained with a K-wire. Occasionally, these distal duplications can be symmetrical raising the question of which thumb to remove. Attempting to preserve attributes of each, Bilhaut²⁷ described a central wedge technique in which the collateral ligaments are saved and only the central



Fig. 46-6 Central polydactyl with a component of syndactyly.

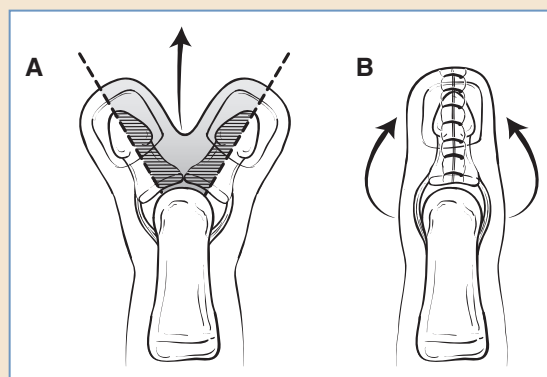


Fig. 46-7 A Bilhaut-Cloquet procedure for symmetrical distal polydactyly. **A**, The preoperative plan. **B**, The postoperative result.

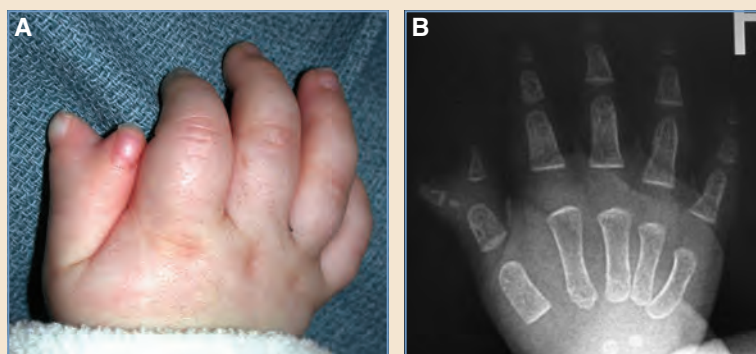


Fig. 46-8 A Wassel type II duplication. **A**, Fully formed distal phalanges. **B**, The radiographic appearance.

redundant tissue is removed (Fig. 46-7). Postoperative grooving of the nail and asymmetrical growth from epiphyseal misalignment plagues this approach.²⁸ Success with a Bilhaut-Cloquet procedure requires precise alignment of the nail matrices and the open epiphyses. This precision is complicated by the lack of gross landmarks confirming the alignment. Consequently, some see no utility for this approach, because the complications are too high. Type II duplications have a full distal phalanx and aberrant flexor/extensor tendon insertions that are often displaced laterally (Fig. 46-8). These must be combined or reattached during the combination procedure bringing their insertion and pull closer to the longitudinal axis of the new digit. Occasionally, the eccentric tendon insertion must be split, creating a Y-shaped insertion that provides a more symmetrical line of pull on the distal bone. Similarly, collateral ligaments need to be reattached if one duplicate is completely removed.

Wassel Types III and IV

The most common type of thumb duplication is Wassel type IV. The common approach to these duplications includes a zigzag incision with a racquet-shaped excision of the thumb to be ablated, which is most often the radial component. Specific problems with this type of duplication include

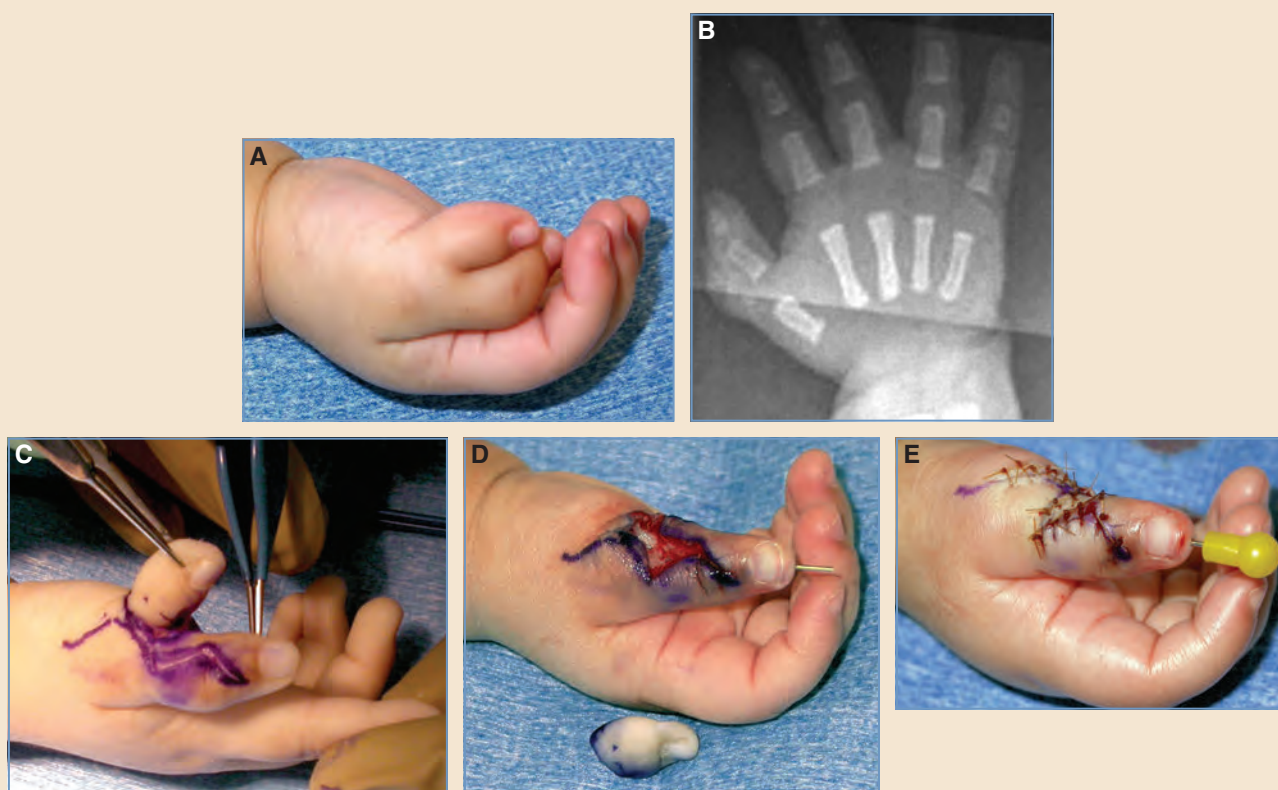


Fig. 46-9 Wassel type IV polydactyly. **A**, Asymmetrical duplication at the metacarpophalangeal joint. **B**, The radiographic appearance. **C**, A zigzag incision with a racquet-shaped excision. **D**, Care in tendinous and ligamentous alignment. **E**, Flap closure and K-wire fixation.

a potentially eccentric insertion of the flexor and extensor tendon apparatus, a wide metacarpal head, lax collateral ligaments, and insertion of the abductor pollicis brevis on the radial thumb. Retaining the ulnar digit is preferable to provide stability at the ulnar collateral ligament for thumb-index pinch. This digit is almost always the larger and more functional of the two. In the rare instance in which this is not so, the surgeon must very carefully reconstruct the ulnar collateral ligament to prevent joint instability. The surgeon can sacrifice joint mobility in favor of stability. The approach may be through a proximally based flap involving the digit to be ablated or through a racquet-type incision at the base of the digit to be ablated, with zigzag skin flap extensions proximally. The flexor and extensor tendons are assessed and their course and insertions centralized and balanced. This is imperative to allow normal motion of the centralized reconstructed digit. The abductor pollicis insertion is dissected off the radial thumb and the radial collateral ligament, maintaining it on a proximally based periosteal flap. It is reinserted on the radial aspect of the ulnar (remaining) proximal phalanx to form a new, secure muscle insertion. The dissection should not be too deep, because the cartilage may be elevated from the epiphysis, inducing future bone growth. After reinserting the tendon and collateral ligament, the surgeon checks the adjustment using a passive tenodesis and thumb-index opposition and secures it with a small axial K-wire to maintain its proper position (Fig. 46-9). If an angulation persists at the interphalangeal joint after correction at the metacarpophalangeal joint, then a closing wedge

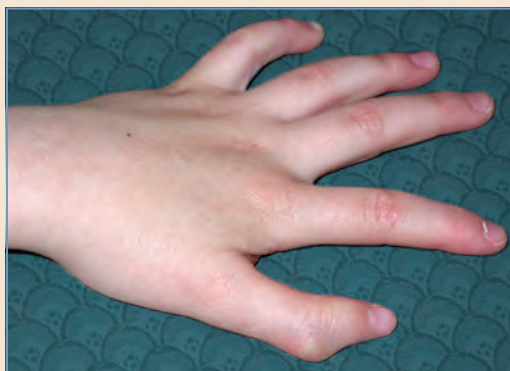


Fig. 46-10 Late angulation from inadequate collateral ligament reconstruction.

osteotomy is required at the distal portion of the proximal phalanx. A bulky dressing and an above-elbow thumb spica-type cast are placed to protect the repair. In type IV polydactyly, the metacarpal head may be excessively wide, and it is tempting to shave off a portion to decrease its width. Although this maneuver actually helps in the ligamentous repositioning of the digit, it creates the potential for growth irregularities. Reduction of the growth plate on one side will reduce the longitudinal growth potential on that side. This may lead to an angular deformity toward the scarred side of the growth plate.

More proximal duplications may have a narrowed first web space that requires opening with a four-flap Z-plasty. Preserving the ulnar collateral ligament prevents late instability and deformity (Fig. 46-10).

Wassel Types V and VI

Although less common than type IV, Wassel type V and type VI duplications must be approached with the same fundamental principles in mind. The duplicated metacarpal has significant attachments from the opponens pollicis and first dorsal interosseous, which are asymmetrical by definition. This anatomy leads to a widened joint surface and nonaxial line of tension. The ulnar duplicated metacarpal lies within the first web space, thus narrowing its overall span.⁷ If symmetrical, excising the ulnar duplicate and reconstructing the ulnar collateral ligament effectively enlarges the first web space without an additional procedure. The extensor mechanism is a combination of extensor pollicis longus ulnarly and extensor pollicis brevis radially. Combining or rebalancing these insertions on the remaining thumb is essential to create an axial line of action. These insertions can be preserved on a small periosteal cuff removed from the excised digit for ease of transfer to the remaining digit.

Triphalangeal Thumb

A TPT can occur independently or as variants of Wassel type IV or VII (see Figs. 46-1 and 46-2). A TPT is often associated with syndromic changes and should prompt a thorough evaluation. An extra middle phalanx is always present with TPT. Early radiographs may not show the cartilaginous middle phalanx, and an apparently wide joint space may be the only clue to its presence. If a duplication is associated with the TPT, the same selection criteria apply regarding which digit will be ablated (that is, the smaller, less stable, less functional digit, which is usually the radial).

However, TPT is difficult to optimize, and this has led some to advocate extirpation of the TPT. Some have proposed transferring the duplicated biphalangeal thumb onto the metacarpal, thereby accepting a shorter digit for increased stability.²⁹

Some surgeons advocate fusing the extra phalanx to the distal or proximal phalanx. This maneuver improves stability and normalizes the joint sequence. An unstable delta phalanx may be best dealt with by excision. Skeletal immaturity makes the radiographic evaluation of this aberrant phalanx difficult, thus complicating the surgical decision-making and postoperative assessment. Multiple reconstructive techniques have been proposed, which in itself is a testament to the difficulty of this reconstruction.^{30,31} For hands with five digits, including a TPT, pollicization is often advised.³²

For all types of thumb duplication, surgeons must attempt to address all of the issues leading to imbalance in the initial operation. The skin, tendons, ligaments, joint surface, and bone should be corrected into axial alignment. The same principles of reconstruction apply, as discussed earlier in the chapter, including tendon balancing, adequate collateral ligament reconstruction, and correction of all angular deforming forces. The impact on the thenar muscle is varied and can be considerable. This muscle deficiency affects the ability of the retained thumb to oppose, regardless of reconstruction technique. Residual asymmetry will not correct itself with time, but will worsen with growth. This creates a curved or zigzag digit that is less functional than a balanced one.

Adjunctive Procedures

With the dual goal of maximizing function and cosmetic appearance, treatment of digital duplication often requires multiple, staged operations. This should be discussed early with parents to prepare them for the overall timeline of correction of this complex problem. The need for revisions to improve stiffness and alignment is common with ongoing development.^{21,22} These procedures include scar revision, corrective osteotomy, arthrodesis, centralization, and collateral ligament reconstruction. Appropriate growth and development should be allowed before initiating revision. However, most abnormalities will worsen, not improve, with time.

POSTOPERATIVE CARE

An ideal dressing is comfortable, immobilizing, and secure. Crafting a bulky dressing that will not slip off challenges even the most experienced surgeons. Incorporating an above-elbow cast often ensures some resistance to the child's attempts to remove it. A dressing change scheduled after 2 weeks with suture removal and reapplication of the splint or cast may require giving a short-acting general anesthetic. The K-wires stay in place for 4 to 6 weeks to allow adequate healing of the joint capsule, collateral ligaments, and osteotomies.²¹ Cast removal and therapy through spontaneous motion begins at 6 weeks. Follow-up evaluation of cosmesis, range of motion, and stability may begin at 3 months. Radiographs should be obtained at this time to assess bony union, bony alignment, and epiphyseal growth potential.

TREATMENT OUTCOMES AND COMPLICATIONS

The success of a congenital hand reconstruction can only be adequately assessed after growth is complete. All parents want their child to have an excellent cosmetic and functional correction of polydactyly. However, this level of restoration is the exception rather than the rule. Ulnar type B pedunculated supernumerary digits are often excised without residual consequence. Thoughtful reconstructive approaches for duplicated thumbs have documented complications ranging from 20% to 50%.^{11,28} Even a conceptually simple Bilhaut-Cloquet central wedge technique

often results in poor growth plate alignment, stiffness, and residual nail deformity.²⁸ Because the epiphyses remain open, minor surgical trauma leads to premature closure and angulated growth. Reoperative rates are high. Before the initial operation, extra time should be spent with parents to emphasize the cosmetic imperfections and the need for secondary functional surgery.

The complexity of thumb duplication and its associated reconstruction lead to equally complex postoperative problems. Often a scar contracture can be managed by simple Z-plasty. Inadequate web spaces can be managed using regional flaps or two- and four-flap Z-plasty. As mentioned previously, axial alignment and stability of the reconstructed digit are paramount. In general, angulation does not correct itself with growth and requires careful assessment of its cause. Tendon insertions or alignment may need precise rebalancing after development has revealed imbalance. Similarly, joint stiffness tends not to resolve, but is functionally better than lax instability at both the interphalangeal and the metacarpophalangeal joint. Instability may require collateral ligament tightening, at minimum, and occasionally arthrodesis.

FUTURE DIAGNOSTIC, ASSESSMENT, AND TREATMENT MODALITIES

Future progress in this field will revolve around research into the exact cause of polydactyly and the possibility of intrauterine fetal surgical correction. With a new understanding of causes and the role of genetics, our current classification scheme may need revision, and alternative treatment modalities will evolve.³²

KEY POINTS

- Possible genetic syndromes should be considered.
- Surgeons should observe how a child uses the hand.
- Three-view radiographs (AP, lateral, and oblique) should be obtained.
- All possible outcomes are discussed with the family, including a timeline of treatments.
- Operative treatment involves the following:
 - Skin is preserved for closure.
 - Tendon insertions are balanced.
 - Collateral ligaments are reattached.
 - Osteotomies are performed to reorient the articular surface.
 - Axial alignment and balance must be accurate.
 - A cast is placed postoperatively to prevent removal by the child.
- Residual deformity is assessed after development.
- Secondary corrections are anticipated and planned.

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47

Congenital Hand Anomalies: Overgrowth

E. Patricia Egerszegi



ongenital overgrowth of a digit and part or all of the upper limb is a rather uncommon condition. It can involve single or multiple tissues, and it can be isolated or occur in conjunction with a syndrome. Some of the earliest reports date back to the first half of the nineteenth century.^{1,2} However, little true understanding of this condition was gained from the late 1800s to recent years, when genetic advances emerged.³⁻⁷ Overgrowth is the clinical manifestation of various unrelated pathologic processes with differing causes. This is a probable explanation for this lack of progress. Contributing further to the confusion, various forms of the condition have been given different names; these include nerve territory-oriented macrodactyly,⁸ club finger,⁹ congenital localized gigantism of the hand, partial gigantocheiry, giant fingers,¹⁰ megalodactyly, dactylomegaly, macrodystrophia lipomatosa, gigantomegaly, local gigantism,¹¹ and macrodystrophia lipomatosa progressive.^{12,13}

CLASSIFICATION

Despite the confusion, a number of authors have tried to classify overgrowth of the upper limb (Table 47-1):

- Temtamy and McKusick's classification¹⁴ is based on whether the anomaly is isolated or part of a syndrome.
- De Laurenzi's classification¹⁵ is dependent on the rate of growth.
 - Stable: The malformation is noticeably larger at birth but proportionately constant in growth with respect to the normal parts of the limb.
 - Progressive: Overgrowth appears at or before 2 to 3 years of age and increases progressively during the growth period at a rate greater than those of the unaffected digits.

Table 47-1 Commonly Used Classification Methods for Macroductyly

Authors	Based on	Particular Features
Temtamy and McKusick ^{14,16}	Isolated	True macroductyly: equivalent to Kelikian and Flatt's type I Pseudomacroductyly: bones uninvolved; soft tissue tumors or anomalies (for example, hemangioma, lipoma, or edema from constriction band syndrome)
	Syndromic	Including: Congenital partial gigantism with metacarpal and phalangeal involvement, subdivided into: (a) Segmental hypertrophy: disproportionately progressive growth of all involved tissues (b) Crossed hypertrophy: affecting different areas on both sides of the body (c) Hemihypertrophy: an incidence of 1/14,300 births ¹⁴ ; digits less severely affected ¹⁶ ; can be associated with Von Recklinghausen neurofibromatosis, Klippel-Trenaunay-Weber syndrome, or Kelikian and Flatt's type IV ¹⁶ ; hamartoma (for example, pigmented nevi, hemangioma) and congenital anomalies (for example, cardiac, polydactyly, or cryptorchidism) present in 20% to 30% of patients ¹⁶ ; association with various tumors (for example, renal, adrenal, brain) ¹⁷ Von Recklinghausen's neurofibromatosis: café-au-lait macules; axillary or inguinal freckling; iris Lisch nodules; characteristic osseous lesions; neurofibroma and other typical neural benign or malignant tumors ¹⁸ ; autosomal dominant inheritance ¹⁹ Ollier disease: multiple, skeletal long-bone enchondroma; most cases sporadic, but mild form can be autosomal dominant ²⁰ ; malignant transformation potential of approximately 30% ²¹ Maffucci syndrome: vascular anomalies and enchondroma; not hereditary, phleboliths in approximately 43%; malignant transformation in approximately 16% to 19% ^{14,20} ; risk of other malignancies (for example, ovarian carcinoma, brain glioma) ²² Klippel-Trenaunay-Weber syndrome: hemihypertrophy of a limb, venous dysplasia, and cutaneous vascular malformation; nonhereditary ²⁰ ; manifestations progress with age ²³ Congenital lymphedema: congenital lymphatic insufficiency; autosomal dominant Milroy disease ²⁴ ; can occur in some other syndromes (for example, Noonan, Turner) ²⁴
De Laurenzi ¹⁵	Stable rate of growth Progressive rate of growth of the involved digits	Present at birth Appearing at or before 2 to 3 years of age
Kelikian ⁸ Flatt ⁹	Tissue involvement and manifestation of the disease process	I: gigantism and lipofibromatosis II: gigantism and neurofibromatosis III: gigantism and digital hyperostosis IV: gigantism and hemihypertrophy

- Kelikian⁸ and Flatt⁹ classified according to the tissue involvement and manifestation of the disease process.
 - I: gigantism and lipofibromatosis
 - II: gigantism and neurofibromatosis
 - III: gigantism and digital hyperostosis
 - IV: gigantism and hemihypertrophy

Ongoing genetic research may eventually lead to the development of a new, improved classification system.

PATIENT CHARACTERISTICS

Macrodactyly is a rare congenital hand anomaly, accounting for approximately 0.9% of all congenital hand anomalies.⁹ According to some authors, the incidence is slightly higher in males than in females¹¹; others note a fairly equal sex distribution.⁹ Most cases are sporadic, with no family history of the condition.^{8,10,11,20} Some associated syndromes have a known inheritance pattern (for example, Von Recklinghausen neurofibromatosis, which has an autosomal dominant pattern^{16,19,20,25}); isolated macrodactyly has no known hereditary cause. Although most patients with this malformation have no other associated malformations, syndactyly occurs in 10% of cases^{8,9,26} (Fig. 47-1). Nevertheless, some syndromic forms of overgrowth have recently been shown to be associated with genetic abnormalities. These include the following^{3,7}:

- Proteus syndrome⁷ (associated with AKT1 mutation³)
- Fibroadipose overgrowth (FAO)³
- Hemihyperplasia multiple lipomatosis (HHML)³
- Congenital lipomatous overgrowth–vascular malformations–epidermal nevi–scoliosis/skeletal and spinal (CLOVES) syndrome³
- Megalencephaly–capillary malformation (MCAP) syndrome³

The last four syndromes listed are associated with PIK3CA mutations.³



Fig. 47-1 A patient with macrodactyly and syndactyly.

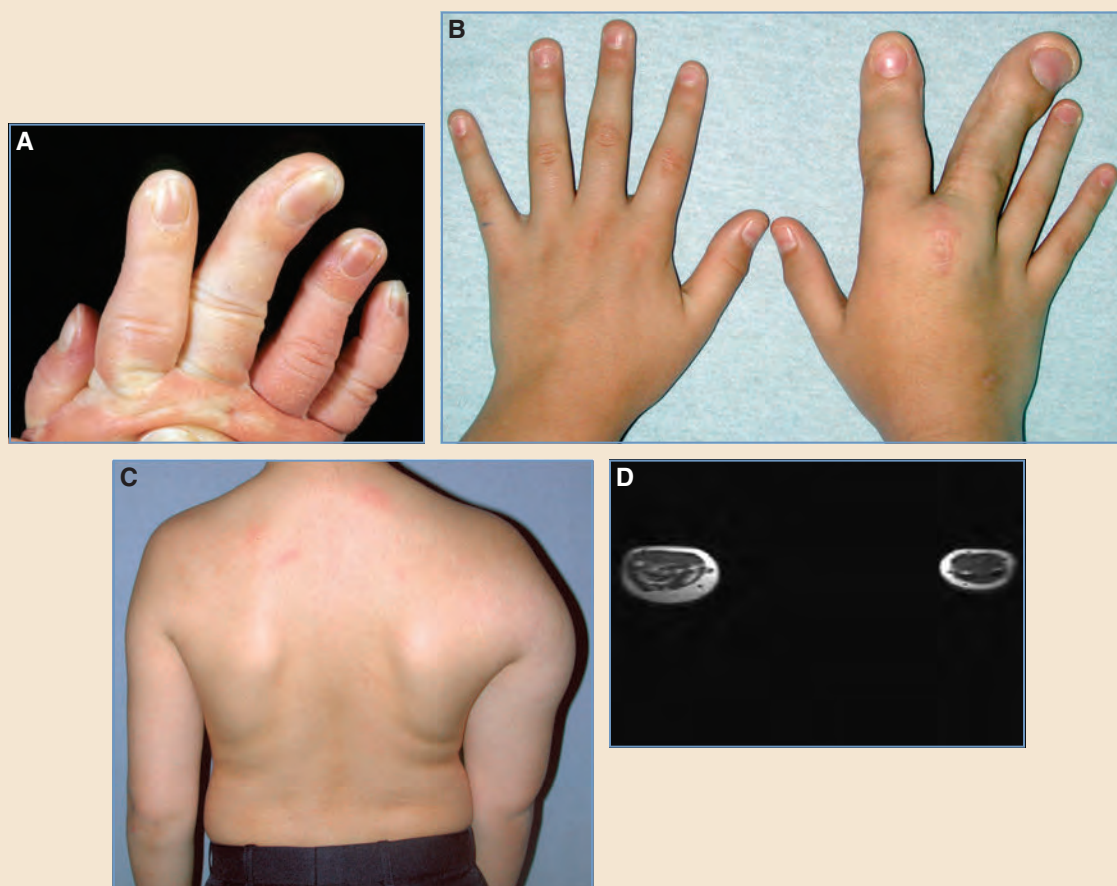


Fig. 47-2 A patient with macrodactyly of the right index and middle fingers and Proteus syndrome. **A**, At 2 months of age, other stigmata of Proteus syndrome are not yet evident. **B**, The patient is shown at 13 years of age after three operations to reduce growth and correct angulation; the clinodactyly has partially recurred. **C** and **D**, Other manifestations of Proteus syndrome are evident at 13 years of age, including a right arm lipoma that was first noticed at 3 years of age. A T1 MRI at the midforearm level shows increased subcutaneous fat in the right upper limb.

Macrodactyly may be one of the earliest presenting features of some syndromic anomalies (Fig. 47-2). Some pediatric tumors can cause enlargement of a digit and should not be confused with macrodactyly.²⁷

HAND CHARACTERISTICS

Approximately 90% to 95% of patients have unilateral hand involvement^{8,9,11,26} (Fig. 47-3). Of these, approximately 60% involve the right hand, although some authors report an equal frequency and others note left-sided predominance.^{3,28} Multiple digits are two to three times more frequently affected than a single one.^{9,11,26,29} In multiple-digit macrodactyly, the involved digits are always adjacent,¹¹ and two digits are more commonly affected than three.^{26,29} When four or more digits are involved, hemihypertrophy is often present and should be evaluated.²⁸ The most commonly involved digit is the index finger (33% to 37%), followed in decreasing order by the middle finger (30%), the thumb (16% to 18%), the ring finger (12% to 14%), and the little finger

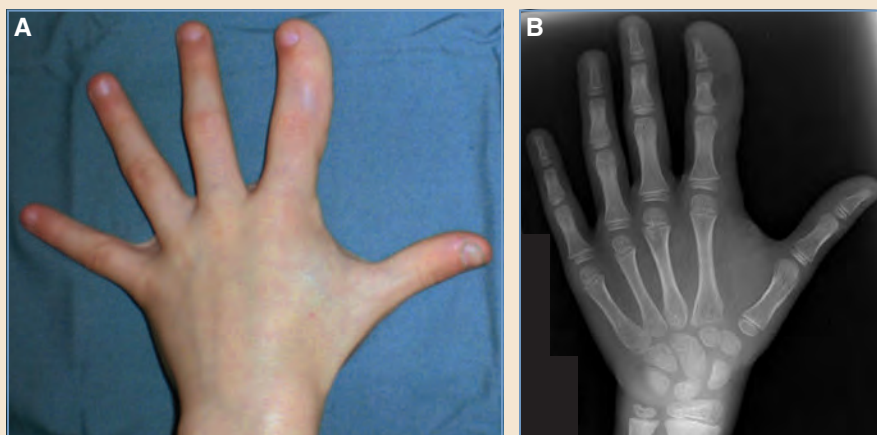


Fig. 47-3 A case involving a single digit. **A**, Single-digit macrodactyly of the left index finger. **B**, A PA radiograph of the left hand. Soft tissue volume is increased on the radial side of the digit.

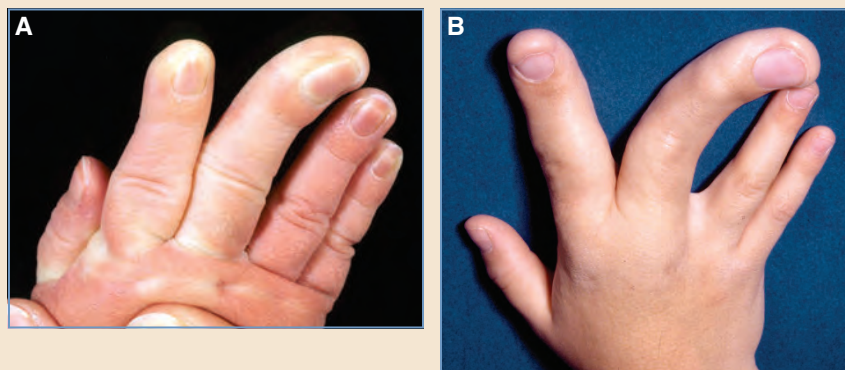


Fig. 47-4 Progression of angular deviation of middle finger macrodactyly. The patient is shown **A**, at 2 months of age and **B**, at 7 years of age.

(3% to 7%).^{11,26} The most typical two-finger combinations are the index-middle and thumb-index, and the least common is the ring-little.^{11,26,29}

Macrodactyly can be limited to a digit or digits, or it can extend proximally various distances along the limb.^{10,11} The enlargement is more pronounced in the distal digit than more proximally.^{9,10,25,30}

The finger is not only longer but also wider than the normal digits.^{25,30} A degree of angular digital deviation in the coronal plane can occur, which can be progressive, forming a curvature (Fig. 47-4). Some authors have reported that the radial finger or fingers deviate ulnarly,^{11,16,28} and others have noted radial deviation.²⁵ The ulnar digits are also noted to deviate radially.^{25,28} Ogino¹¹ specified that the ring finger can deviate in either direction depending on whether the median or ulnar nerve is involved. However, when adjacent fingers are affected, they tend to diverge, according to some authors,^{10,25,28} and converge according to others.⁹ The affected thumb commonly assumes a hyperextended and abducted posture.^{9,16,25} Because the palmar aspect tends to be more severely involved than the dorsal surface,^{25,30} dorsal deviation can occur^{9,16} and can even be pro-



Fig. 47-5 Limited flexion of macrodactylous index and middle finger distal interphalangeal joints.

gressive. The joints often have limited flexion,^{25,30} particularly the distal interphalangeal joint,^{11,30} and this can progress with growth (Fig. 47-5), resulting in functional deterioration.^{9,31} Although longitudinal growth tends to stop with epiphyseal closure, in some cases transverse growth¹⁶ or the soft tissues may continue to increase.³² Pure nerve impairment is uncommon; however, compression neuropathy such as carpal tunnel syndrome can occur and necessitate decompression.^{9,11,33-36}

CAUSE AND PATHOLOGY

Although the exact cause of most types of macrodactyly is unknown, neural,^{9,30,31,37} vascular,³⁷ humoral,^{37,38} genetic,^{3-6,39} and a combination of genetic and environmental factors²⁹ have been proposed as possible causes of this anomaly.

Although some have discussed a relationship between macrodactyly and Von Recklinghausen neurofibromatosis, several authors agree that there are more differences than similarities.^{8,10,11} Many cases of macrodactyly are associated with enlarged nerves in the affected area.^{9-11,30,33,40} This has led to the term *nerve territory-oriented macrodactyly*.⁸ Some have interpreted this association to imply that the nerve is the cause of the disorder,^{9,25,30} because certain nerve disorders are known to affect growth (for example, brachial plexus injury results in decreased limb growth). Nevertheless, it is entirely possible that whatever causes enlargement of the affected structures also affects the nerves.³⁴ Nerve resection does not diminish subsequent growth. This supports the hypothesis that nerve enlargement is unlikely to be the only factor in the overgrowth.¹¹ It has been suggested that molecular biology or genetic studies for somatic mutations may eventually lead to a better understanding of the condition and its causative factors.^{38,41} Recent advances in genetic research have provided evidence that some forms of overgrowth may be related to anomalies at the genetic level, such as pleiotrophin upregulation⁴; mutation of PIK3CA^{3,4}; a gain-of-function mutation of the natriuretic peptide receptor 2 gene, causing excessive production of cGMP^{5,39}; and mutation of AKT1.³ Evidence exists that certain forms of overgrowth are limited to the somatic cell, and that they can arise from the same genetic foundations as some generalized mosaic overgrowth syndromes.⁶ These findings beg the question of whether timing and location of the genetic event may be factors explaining the difference between certain generalized and localized phenotypes.^{3,6}

The pathologic findings seen in macrodactyly include thickening of the skin with decreased sweat gland density.⁴² Increased subcutaneous fat with fibrosis,⁴² similar to that in adults, is found



Fig. 47-6 An enlarged median nerve extending proximally into the carpal tunnel of the right hand in a patient with macrodactyly.

with large, darker, firmly fixed lobules.²⁹ The tendons are generally not involved,²⁹ although they may be increased in size,¹⁰ with a thickened tendon sheath.²⁵ The arteries are enlarged,⁴³ particularly because of increased luminal caliber,¹¹ and, according to Flatt,⁹ during growth may not always keep up with tissue requirements, resulting in vascular insufficiency. The bone tends to be enlarged in all directions, with a widened medullary canal^{9,16,44} and thickened,^{25,44} fibrotic periosteum.^{37,44,45} Bone age is advanced in the involved regions as evidenced by an earlier appearance of the epiphyseal secondary calcification centers.^{10,11} The digital nerves, which are enlarged in width and length, are difficult to separate from the surrounding subcutaneous tissue and can have a tortuous course.^{10,25} Involvement often extends proximally into the hand and beyond^{9,11,16} (Fig. 47-6). There is an increase in fibrous and fatty tissue around the nerve.^{29,44} Some authors have also described thinning or degenerative changes of the myelin, which they interpret as effects of compression.^{30,46}

In the specific case of macrodactyly associated with hyperostosis, osteocartilaginous masses are present around the joints,^{11,25} and nerves may not be enlarged^{9,10,25} or may be infiltrated with fibrofatty tissue.¹⁶ This may therefore be an unrelated entity. Nevertheless, the involved area seems to follow a neural territory.⁹

PATIENT ASSESSMENT

Once a tumor^{27,47} diagnosis has been excluded—by history, examination, investigation, and, when indicated, biopsy—it is important to establish if the patient's macrodactyly is isolated or part of a syndrome. Macrodactyly that is part of a syndrome may require consultation with and treatment from other specialists or an interdisciplinary clinic. Symptoms of compression neuropathy should be sought in patients with significant palmar or proximal involvement, particularly in those old enough to collaborate for a reliable evaluation. Otherwise, patients are assessed for signs of this problem, because significant compression may begin at an early age and require decompression. EMG and nerve conduction studies can be helpful. Evaluation by an occupational therapist can aid in assessing function, identifying deterioration, and establishing the best treatment plan.

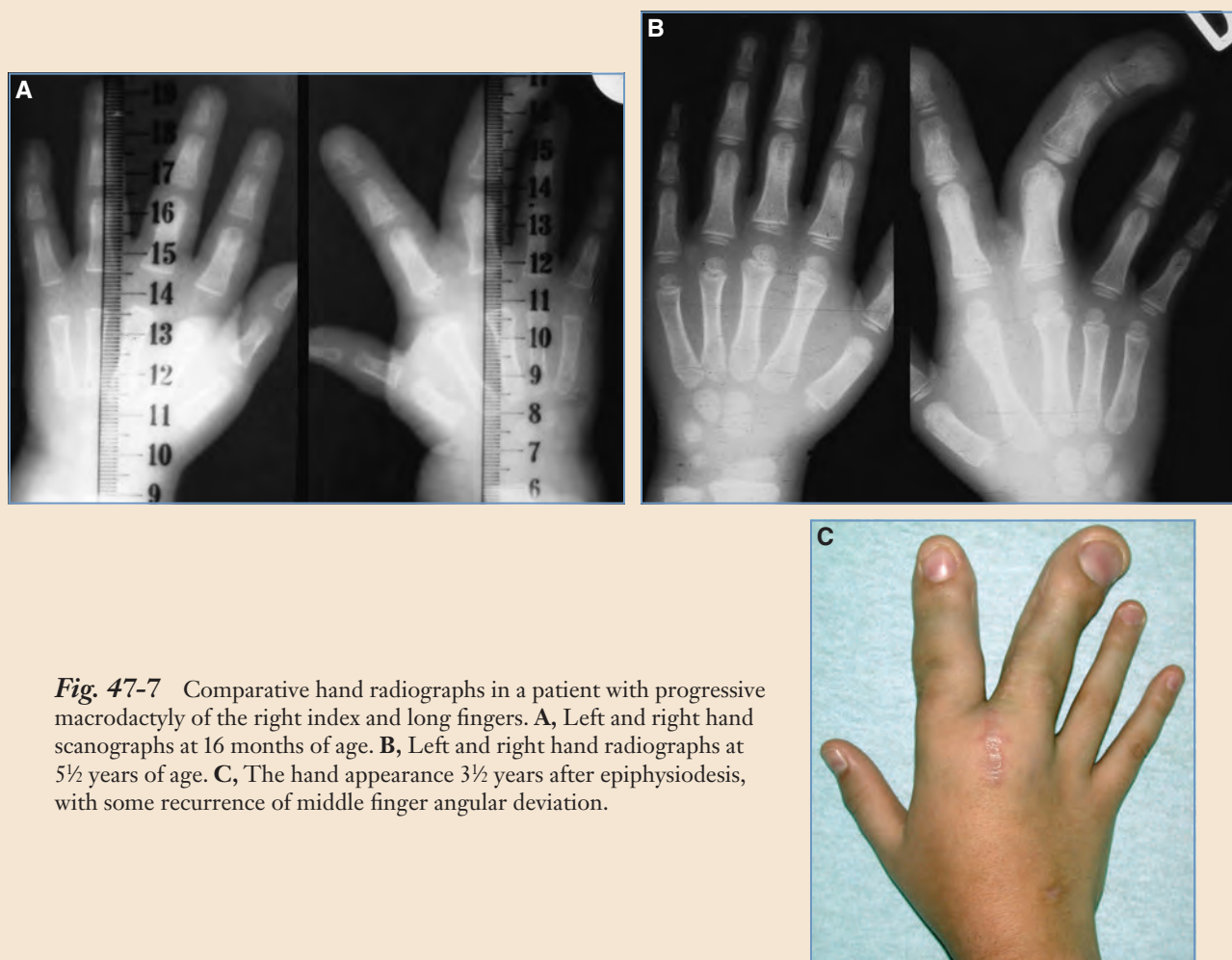


Fig. 47-7 Comparative hand radiographs in a patient with progressive macrodactyly of the right index and long fingers. **A**, Left and right hand scanographs at 16 months of age. **B**, Left and right hand radiographs at 5½ years of age. **C**, The hand appearance 3½ years after epiphysiodesis, with some recurrence of middle finger angular deviation.

Sequential comparative radiographs of the normal and involved hands and/or limbs help to determine whether the condition is static or progressive. These may include, if indicated, scanographs to assess for a limb length discrepancy (Fig. 47-7). Radiographs also are used for measuring the degree of coronal or dorsopalmar deviation and the exact sites contributing to the angulation. In some cases, a psychological evaluation and follow-up support may be useful in deciding the best treatment plan.

TREATMENT OPTIONS

There is no useful nonsurgical treatment of macrodactyly. Epiphyseal plate arrest through microangiography and embolization by ultraspecialized interventional radiologists may one day become a reality. However, this technique's use is limited to the treatment of larger bones, such as the ulna or radius (J. Dubois, personal communication, 2005). Although surgical treatment

of macrodactyly has no formal contraindications, short of life-threatening conditions, surgical treatment is controversial in some cases. These include more subtle forms with no functional impairment of the hand and patients who do not wish to have surgery.

In most cases, surgery must be tailored to each patient's needs, deformity, functional problems, and wishes. Staged surgery is often necessary⁴⁸ and long-term follow-up required.

PREOPERATIVE PLANNING

Historically, surgical treatment is subdivided into four general categories:

1. Early attempts at inhibition of further overgrowth
2. Intermediate-period longitudinal growth arrest
3. Early- to late-period debulking procedures in length and/or width
4. Other corrective procedures, including nerve decompression, correction of angulation deformities, or even amputation

The last category of corrective procedures can be undertaken at any time but is usually reserved for the intermediate and late periods and is not discussed in this chapter.

Surgical Plan

The choice of procedure depends at least in part on these factors:

- The patient's age at presentation
- Whether the malformation is static or progressive
- The severity of the macrodactyly
- Whether the digit has attained adult size
- Whether the curvature is significant
- The range of motion at each joint
- Whether multiple digits are involved

In my experience, and most surgeons agree, nerve stripping or excision does not slow the growth of a digit, even in young patients.^{9,49} However, nerve excision can be useful during otherwise planned debulking procedures, and, if undertaken bilaterally in a specific digit, at least one should be reconstructed with a nerve graft.¹¹ Because vascularization in these fingers is frequently precarious,⁹ ideally both or at least one digital artery should be preserved during nerve excision. If both sides of a finger require treatment, 2 to 6 months between procedures is suggested (depending on the author and technique).^{9,10,16,28,49-51} This may decrease the likelihood of vascular compromise.

To prevent longitudinal overgrowth of a finger if the growth plates are still open and the digit has grown to adult length, epiphysiodesis is indicated at the involved levels (usually all phalanges, possibly including the metacarpal). Angulation deformities can be corrected simultaneously. The incisions used should not interfere with further planned surgery.

Many previously described combination techniques for digit reduction correct longitudinal bony and soft tissue overgrowth. However, they generally address only soft tissue reduction in width. When treated using these techniques, grossly enlarged digits that are longer than adult size are shortened but often remain disproportionately wide. Although only a case report, the technique reported by Bertelli et al⁵⁰ shows promise, because it addresses both bony and soft tissue width and length. The technique includes a first operation involving resection of the ulnar thirds of the phalanges, detachment and reinsertion of the proximal interphalangeal joint collateral ligament, resection and fusion of the distal interphalangeal joint, removal of the ulnar soft tissues, including the neurovascular pedicle on the same side, and excision of the distal third of



Fig. 47-8 Amputation of a macrodactylous digit. **A**, This patient had an epiphysiodesis of the phalanges and soft tissue debulking of the middle finger of the left hand at 28 months of age. The patient is shown 3 years postoperatively. **B**, The middle finger was amputated when the patient was 5½ years of age. An epiphysiodesis combined with closing wedge osteotomies of the phalanges with soft tissue debulking of the ring finger was completed at 9 years of age. The patient is shown at 12 years of age.

the nail bed. Six months later, debulking is undertaken on the radial side, preserving the radial neurovascular pedicle. Although the result is impressive, the technique's worth needs to be confirmed in a greater number of patients.

Finally, for some grossly enlarged digits that obstruct function, amputation may be the best treatment option (Fig. 47-8).

Positioning and Marking

The patient is placed in a supine position, with the involved limb abducted and resting on a hand table. In very small patients, the head and shoulder can rest on the most proximal part of a sufficiently wide hand table. The rest of the body is positioned slightly obliquely away from the hand table toward the opposite-side foot of the main operating room table, because this allows the hand to extend farther onto the hand table, facilitating the surgeon's approach to the hand (Fig. 47-9). A tourniquet is applied and used intraoperatively (inflated to 200 mm Hg or less in younger children and at 250 mm Hg or less in older, healthy, normotensive children).^{52,53} The tourniquet is deflated for at least 15 minutes⁵³ after 1½ to 2 hours of inflation, and then it is reinflated.^{52,53} However, according to Crenshaw,⁵² it is less traumatizing to leave the tourniquet on for 2 hours or even a few minutes longer, then deflate and not reinflate it, than to deflate for 10 minutes and reinflate for the necessary additional time. Markings are made according to the technique planned (as described later in the chapter), and the patient is prepared and draped as usual. Prophylactic antibiotic agents are generally not given unless specifically indicated (for example, for multiple episodes of ingrown nail or prolonged surgery).



Fig. 47-9 Positioning a small child for hand surgery to allow the hand to lie far enough distally on the hand table to facilitate the surgeon's work. The head lies partly on the hand table and the body obliquely on the operating room table. The shoulder should not be abducted much beyond 90 degrees, and the head should not be completely turned to the opposite side to prevent traction on the brachial plexus.

SURGICAL TECHNIQUE

Early Inhibition of Further Overgrowth

Some authors have claimed that complete or partial digital nerve resection or stripping of its branches can prevent digital overgrowth.^{30,32} In my experience, and it is now generally agreed, this does not slow or stop growth.^{9,49} Nevertheless, if considered in conjunction with a debulking procedure, for example, it should be performed early enough to allow some reinnervation after nerve excision.¹¹ Nerve procedures may include excision of the hypertrophied part of the nerve and reanastomosis^{8,10,11}; nerve excision, with or without the skin it innervates^{11,16,49} (if both digital nerves are excised, reconstruction of one using a nerve graft is recommended¹¹); or longitudinal partial median nerve resection.⁵⁴ This last technique may be difficult or impossible if the nerve fascicles are torturous. Efforts must be made to keep the digital arteries intact⁴⁹ to minimize the chance of vascular compromise. The incisions are usually made according to the debulking procedure performed and the future planned surgery. If both sides of a finger require treatment, a period of 2 to 6 months between procedures (depending on both the author and the technique) is suggested.^{9,10,16,28,49-51}

Intermediate-Period Longitudinal Growth Arrest

Most authors agree that longitudinal growth should be arrested when the digit is the same length as the corresponding digit in the parent of the same sex. According to Ogino,¹¹ surgery is usually performed when patients are about 10 years of age. In larger bones (for example, the radius

and ulna), the use of intraarterial embolization of the growth plate may be possible (J. Dubois, personal communication, 2005). However, in small bones, such as those in the hand, such techniques have not been used for fear of compromising the distal circulation of the digit. Surgical epiphysiodesis is therefore the mainstay of longitudinal growth arrest. Various methods have been described to stop longitudinal bone growth, including cauterization,⁵¹ high-speed burring,⁵¹ surgical curettage,^{51,55} and surgical excision.⁵⁵ Although stapling has been attempted, it has proven too difficult to position the small staples across the epiphyseal plate.^{9,56} Angular deformity sometimes occurs after epiphyseal arrest (Fig. 47-10), and may require surgical treatment with a wedge osteotomy.⁴³ If a degree of deviation is present in any plane when epiphysiodesis is undertaken, a wedge osteotomy can be performed simultaneously^{25,49} (Fig. 47-11). Some have attempted hemiepiphysiodesis on the convex side of the finger, but results have been less reliable than those obtained with a corrective osteotomy.²⁵



Fig. 47-10 Recurrence of angular deviation after a closing wedge osteotomy followed by epiphysiodesis of the macrodactylous middle finger phalanges. **A**, The hand appearance before bony intervention. **B**, The hand appearance 2½ months after an epiphysiodesis of the index and middle finger phalanges and closing wedge osteotomies of the middle finger proximal and middle phalanges. **C**, The hand is shown 3½ years after an epiphysiodesis, with some recurrence of middle finger angular deviation.



Fig. 47-11 A combination of epiphysiodesis and closing wedge osteotomies to correct angulation and arrest longitudinal growth. **A**, A radiograph obtained 1 year after closing wedge osteotomies at the junction of the middle and distal thirds of both the proximal and middle phalanges of the middle finger and before an epiphysiodesis. **B**, A radiograph obtained 16 months after an epiphysiodesis of all phalanges of the index and middle fingers, including closing wedge osteotomies at the level of the epiphysiodesis for the proximal phalanx of the index finger and both proximal and distal phalanges of the middle finger.

Although surgeons should take into account future surgeries when planning cutaneous incisions, they are often made in the midlateral line on the convex side of the digit.

Early- to Late-Period Debulking Procedures

Early- to late-period debulking procedures include those aimed at the soft tissues, the bone, or both. Soft tissue procedures can be undertaken at any time and involve variable and often sequential excisions of skin and subcutaneous tissue, including or excluding the digital nerve or its branches. Most surgeons recommend debulking half the circumference at a time, with an interval of 2 to 6 months between procedures^{9,10,16,28,49-51}; however, a palmar pedicled flap and a dorsal island flap have been described, one based on the radial and the other on the ulnar neurovascular digital pedicle.⁵⁸ It is important to preserve the digital arteries, particularly in cases in which debulking is planned bilaterally.⁴⁹ Most authors describe using a midlateral incision.^{9,25,28,49} Some authors have mentioned a Brunner incision,^{9,16,25,50} but Flatt⁹ has reported that the resulting scars are more frequently hypertrophic.

Bony reduction procedures tend to be reserved for the intermediate and late periods. They include reductions in length using resection osteotomies to shorten bones,^{11,16,25} which can be combined with a correction of angular deviation^{9,16,25} and/or epiphyseal arrest.¹⁶ Shortening can be accomplished using a partial or complete digital metacarpophalangeal joint resection,⁴⁹ an arthrodesis (particularly for the thumb metacarpophalangeal joint, as long as the carpometacarpal and interphalangeal joints have satisfactory motion),^{9,11,25,28} and a distal interphalangeal joint excision and arthrodesis in the digits.^{11,29,50,57-59} Complete excision of the middle phalanx⁶⁰ has been proposed for severe problems.^{9,16,25,28} Similarly, terminalization (amputation of the distal phalanx, including the nail) has also been described, but is generally not recommended in the hand for cosmetic reasons.^{9,16,25,28} Other procedures include resection of the distal phalanx and the distal part of the middle phalanx, preserving the nail complex as a pedicled island flap,⁶¹ and a partial distal phalangeal tip reduction,⁴⁹ which is a component in several authors' combination techniques.^{49,59,62-64}

Decreasing bony width by resecting a quarter to a third of each side of each phalanx⁹ is thought to reduce articular motion and should probably be reserved for cases in which motion is already greatly reduced, in periarticular areas where arthrodesis is planned, or for cases in which aesthetic considerations outweigh functional ones. Nevertheless, Bertelli et al⁵⁰ reported a case (described previously) of unilateral longitudinal resection of a third of the bony width of the phalanges in an 8-year-old girl in whom metacarpophalangeal and proximal interphalangeal joint motion remained unchanged after 4 years of follow-up. An epiphysiodesis was not undertaken, with the exception of that of the distal phalanx at the time of an arthrodesis of the distal interphalangeal joint. This procedure is worrisome to undertake in young patients with little or no joint stiffness; however, it is not known whether such a surgery would provide better results than those obtained when patients are older; in my experience, ligament reconstruction is superior (less instability and less stiffness) in younger patients (12 to 24 months of age) for other types of congenital hand malformations (for example, thumb duplication). Inspired by the procedure of Bertelli et al, Kakinoki and Ikeguchi⁵⁸ performed an even more aggressive operation in even younger patients (3 years of age). The bony procedures were similar, only with greater shortening of the distal part of the middle phalanx and reduction in the width of the proximal phalanx extending more proximally. The bony reconstruction was then covered using a palmar pedicled and a dorsal island flap, one based on the radial and the other on the ulnar neurovascular digital pedicles. The dorsal flap also included dorsal subcutaneous veins. Despite such extensive surgery, metacarpophalangeal joint motion remained stable, and proximal interphalangeal joint motion actually increased from 40 degrees to 70 degrees at the 3-year follow-up.

Most debulking techniques combine several procedures simultaneously and/or sequentially, often including longitudinal growth arrest or various modalities to correct angulation deformities. Although multiple methods have been described, only Millesi's thumb reduction method⁶⁴ (see Fig. 47-12) and the digital shortening techniques of the following authors will be described and illustrated in this chapter, including the necessary incisions: Barsky²⁹ (see Fig. 47-13), Tsuge⁴⁹ (who now limits his frequently cited 1967 operation³⁰ to treatment of the toes) (see Fig. 47-14), Hoshi et al⁵⁹ (see Fig. 47-15), Fujita et al⁶³ (see Fig. 47-16), Ogino¹¹ (see Fig. 47-17), and Bertelli et al⁵⁰ (see Fig. 47-18).

Millesi's Thumb Reduction Method

Millesi's thumb reduction method⁶⁴ includes resection of the distal phalanx distally and along a central longitudinal strip and shortening of the proximal phalanx using an oblique resection osteotomy (Fig. 47-12). The reduction in width of the distal phalanx is limited by the interphalangeal joint's collateral ligament laxity; the proximal phalanx width is not addressed. Because the distal phalanx osteotomy involves the joint surface, any remaining motion may be limited. A nail deformity in the form of a ridge or a separation often occurs, because soft tissue excision includes the nail bed and root in the midline. An extensor lag may result, which requires tendon shortening. In this case, the tendon may adhere to the underlying osteotomy of the proximal phalanx, further limiting motion.

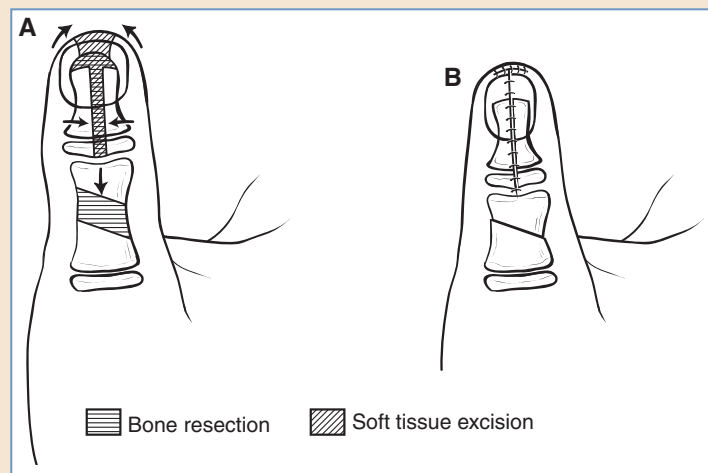


Fig. 47-12 Millesi's thumb reduction method. **A**, The surgical plan (dorsal view). The distal phalanx is resected distally and along a central longitudinal strip. The strip width is limited by the interphalangeal joint's collateral ligament laxity. Internal fixation can be undertaken using an interosseous wire.¹⁶ The proximal phalanx is shortened using an oblique resection osteotomy. Internal fixation can be accomplished using an interosseous wire and a K-wire perpendicular to the osteotomy.¹⁶ The approach can be through a longitudinal midline incision. Soft tissue excision includes the nail bed and root. The fingertip soft tissue is closed by rotating radial and ulnar skin flaps toward each other. **B**, The postoperative appearance.

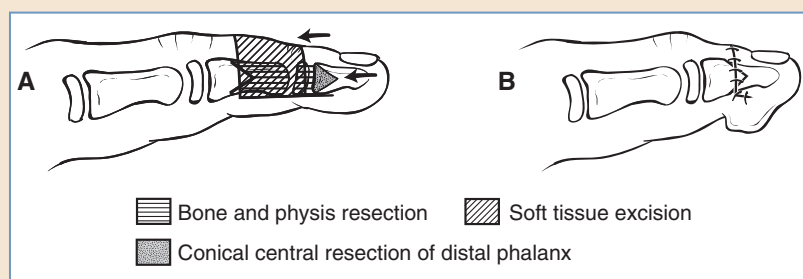


Fig. 47-13 Barsky's procedure. **A**, The surgical plan (lateral view). Bone resection includes the distal part of the middle phalanx, the distal phalanx articular surface, and a conical distal phalanx central core, including all or much of the growth plate, to receive the sharpened end of the middle phalanx. Internal fixation can be achieved using a longitudinal K-wire. The extensor tendon is shortened and the deep flexor tendon plicated. Six weeks postoperatively, excess palmar soft tissue can be excised. In a simplification of this technique, transverse osteotomies facilitate resection of the distal interphalangeal joint.¹⁶ (Although this modification is attributed to Flatt,⁹ it does not appear in the cited reference.) **B**, The postoperative appearance.

Barsky

Barsky's procedure²⁹ involves resection of the distal part of the middle phalanx, the distal phalanx articular surface, and the conical distal phalanx central core, including all or much of the growth plate, to receive the sharpened end of the middle phalanx (Fig. 47-13). The extensor tendon is shortened and the deep flexor tendon plicated. Excess palmar soft tissue can be excised 6 weeks postoperatively. Although this technique shortens the longitudinal dimension of the finger to a certain extent, it does not reduce the width, and any motion present at the distal interphalangeal joint is lost. This technique does not address continued proximal or middle phalangeal growth or angulation deformities.

Tsuge

In Tsuge's 1985 operation⁴⁹ the phalangeal growth plates are excised and a tip plasty, including excision of bony and soft tissues, is performed with or without digital soft tissue and unilateral digital nerve resection (Fig. 47-14). Further bone shortening is carried out as needed, beginning with the base of the proximal phalanx, and, if required, the head of the metacarpal. According to Tsuge, in older children, the metacarpal growth plate is left intact to prevent joint stiffness. He did not specify what is done to the metacarpophalangeal collateral ligaments. Using this method, not only is further growth reduced, but length is also significantly reduced, although potentially at the expense of metacarpophalangeal joint stability or motion. On the other hand, the reduction in the width of the digit is more modest, because reduction of the bony contribution to this dimension is not attempted. Although not formally part of the description, this technique could be used potentially to correct proximal angulation deformities but not those located more distally, by including a wedge osteotomy to achieve epiphysiodesis. A resulting extensor lag in cases with more aggressive reduction in length would not be surprising and would require tendon shortening. Tendon adhesions to the operated metacarpophalangeal area may develop in some such cases.

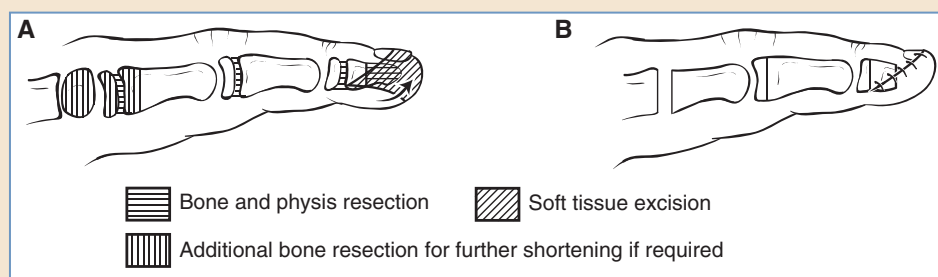


Fig. 47-14 Tsuge's operation. **A**, The surgical plan (lateral view). The phalangeal growth plates are excised, along with bone and soft tissue at the tip of the finger. Additional bone is resected for further shortening, if needed, through a dorsal midline incision, beginning with the base of the proximal phalanx and, if required, the head of the metacarpal. In older children, the growth plate is preserved intact to prevent joint stiffness. Tsuge's article does not specify what is done to the metacarpophalangeal collateral ligaments. Soft tissue resection may be continued proximally in the midlateral line to include hypertrophic nerve and excess skin if debulking is required. **B**, The postoperative appearance.

Originally proposed by Tsuge,³⁰ complete resection of a digital nerve is only indicated in selected cases and usually limited to a single side. Although several authors have reported that patients with macrodactyly do not have a significant reduction in sensation,^{49,50,62} no systematic, double blind, complete, formal evaluation of patients having this procedure has ever been conducted. In my experience, nerve excision does not slow or halt growth and should not be used for this purpose.^{9,49}

Hoshi et al

The technique described by Hoshi et al⁵⁹ consists of a resection osteotomy of the proximal metacarpal, resection of the distal end of the terminal phalanx, and resection and fusion of the distal interphalangeal joint (Fig. 47-15). Soft tissue is excised on the convex side of the digit, including the distal and convex sides of the nail bed and root. The reduction in length is achieved at the expense of the distal phalanx and distal interphalangeal joint, similar to Barsky's operation,²⁹ in addition to further shortening at the metacarpal level. As with Barsky's technique, disadvantages include loss of any motion present at the distal interphalangeal joint. In addition, this technique does not address continued growth of the proximal or middle phalanx or any angulation deformities. On the other hand, there is more reduction in width than with Barsky's operation because of the soft tissue reduction on the convex side of the digit and nail. Reduction in width remains more modest than if a bony procedure had also been included.

Fujita et al

In the shortening method described by Fujita et al,⁶³ length reduction is limited to bone resection of the distal end of the terminal phalanx and excision of the distal dorsal and palmar soft tissues (Fig. 47-16). Width is reduced by removing soft tissue. A proximally based palmar soft tissue flap is created, including the radial neurovascular pedicle. The ulnar third of the flap and the ulnar digital nerve are excised. Reductions in length and width are more modest, because bony reduction in length is limited to one level, and bony width is not reduced. This procedure does not limit continued growth or correct angulation deformities. Once the ulnar digital nerve has been dissected away from the artery and excised, the vascular supply to the dorsal skin and bony

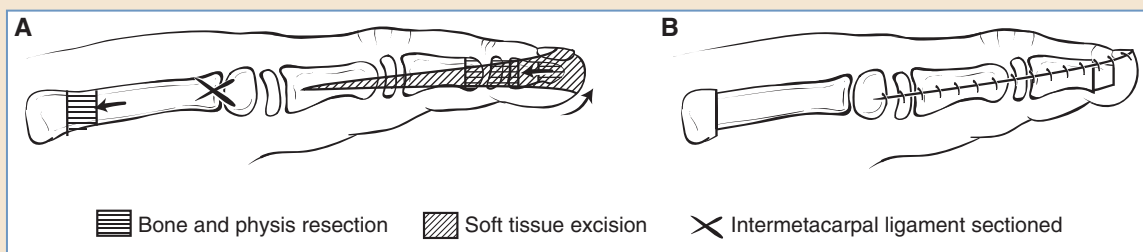


Fig. 47-15 The technique of Hoshi et al. **A**, The surgical plan (lateral view, convex side). Bone resection includes a resection osteotomy of the proximal metacarpal, which can be performed through a dorsal longitudinal incision; resection of the distal end of the terminal phalanx; and resection and fusion of the distal interphalangeal joint, performed through the soft tissue excision sites. Bone is fixated using a K-wire and/or interosseous wire. The intermetacarpal ligament or ligaments must be transected to allow shortening of the digit. Soft tissue excision is performed on the convex side of the digit, including the distal and convex side of the nail bed and root. **B**, The postoperative appearance.

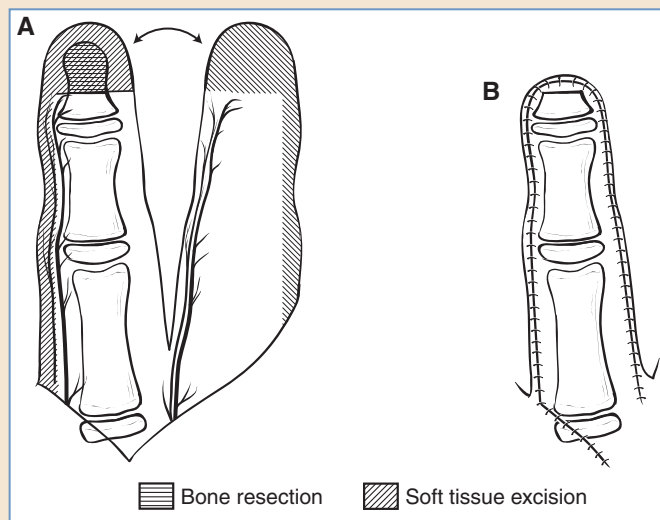


Fig. 47-16 The method of Fujita et al. **A**, The surgical plan (palmar view). Bone resection is limited to the distal end of the terminal phalanx. The proximally based palmar soft tissue flap includes the radial neurovascular pedicle. The distal end and ulnar third of the flap are excised, along with the ulnar digital nerve. The tip is closed with the distal end of the flap. **B**, The postoperative appearance.

skeleton, and even to the palmar flap, may be a concern. Dorsal and ulnar digital sensation merits study. In my experience, and as Flatt⁹ reported, scarring is often hypertrophic in these patients. This may become problematic because of the extent of the incisions.

Ogino

Ogino's procedure¹¹ involves bone resection of the proximal phalanx, including a closing wedge osteotomy to correct angulation, resection of the distal end of the terminal phalanx, and resection and fusion of the distal interphalangeal joint (Fig. 47-17). Soft tissue is excised on the convex side and includes the digital nerve but excludes the digital artery. Similar to the operations described by Barsky²⁹ and Hoshi et al,⁵⁹ the length is reduced at the expense of the distal phalanx and distal interphalangeal joint, and by further shortening at the proximal phalangeal level. This technique

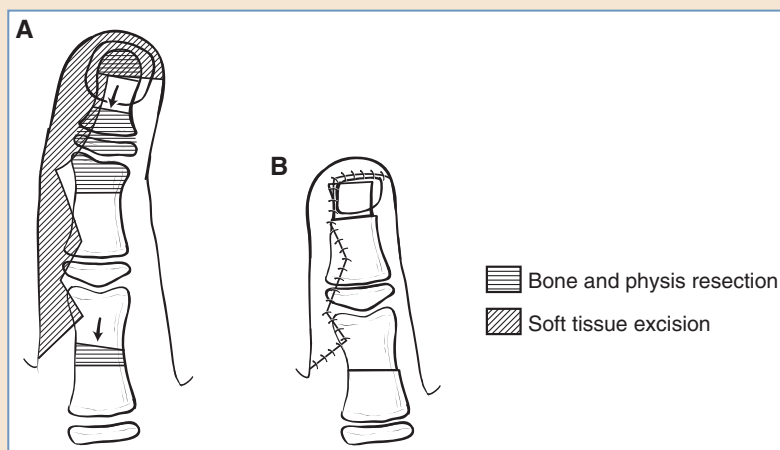


Fig. 47-17 Ogino's procedure. **A**, Surgical plan (dorsal view). Bone resection includes a part of the proximal phalanx and a closing wedge osteotomy to correct angulation, resection of the distal end of the terminal phalanx (which is mentioned in the text, not included in the reference diagrams, but added here), and resection and fusion of the distal interphalangeal joint. Internal fixation can be achieved using K-wires and/or interosseous wires. Soft tissue excision is undertaken on the convex side, including the digital nerve but excluding the digital artery, and closed with interdigitating palmar and dorsal flaps. **B**, The postoperative appearance.

includes the correction of an angulation deformity. As with the techniques of Barsky and Hoshi et al, disadvantages include loss of motion at the distal interphalangeal joint and failure to address proximal or middle phalangeal continued growth. Because of shortening at the proximal phalangeal level, an extensor lag may result, which may require tendon shortening. In this case, the tendon may adhere to the underlying osteotomy of the proximal phalanx, further limiting motion. Sensation on the side of the resected digital nerve merits study.

Bertelli et al

As described previously, Bertelli et al⁵⁰ have performed an operation with the potential advantage of correcting both bony and soft tissue excess in width (Fig. 47-18). Similar to Barsky's procedure,²⁹ bony length reduction is more modest and is limited to shortening at the level of the distal interphalangeal arthrodesis. As with Barsky's, Hoshi's, and Ogino's techniques,^{11,29,59} disadvantages include loss of any motion present at the distal interphalangeal joint, and proximal or middle phalangeal continued growth is not addressed. In addition, the potential for stiffness or instability exists at the proximal interphalangeal joint collateral ligament reinsertion site. This operation does treat angulation deformities. The vascular supply can be a concern, because one neurovascular pedicle is completely resected. As in Tsuge's and Fujita's methods,^{49,63} sensation after complete excision of one digital nerve would merit further study.

In summary, the most aggressive reduction in width is achieved with the operation described by Bertelli et al,⁵⁰ and the techniques of Hoshi et al⁵⁹ and Ogino¹¹ produce the greatest osseous shortening. Epiphysiodesis can be combined with most techniques once the digit is the same length as that of the parent of the same sex. Angulation deformity can often be corrected at the same time as the epiphysiodesis by incorporating a closing wedge osteotomy. The most aggres-

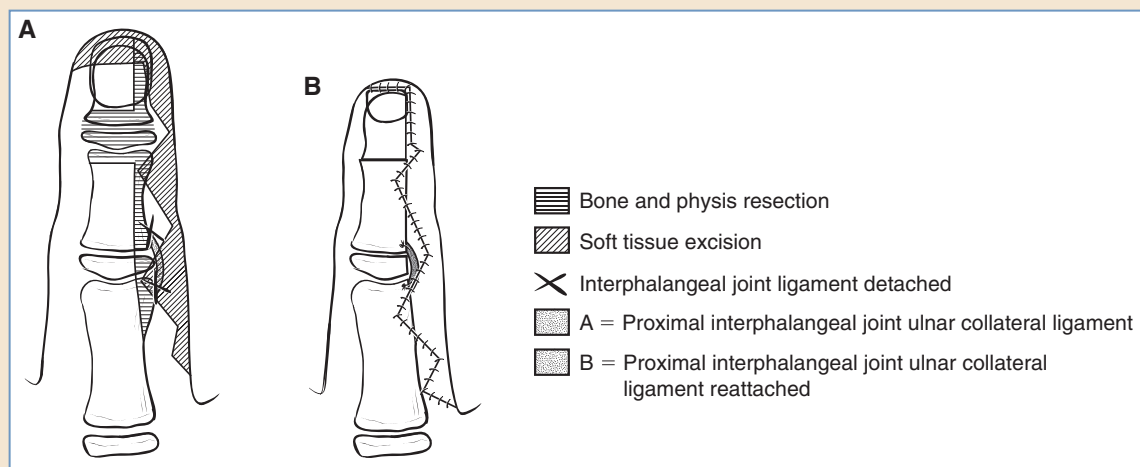


Fig. 47-18 The operation of Bertelli et al. **A**, The surgical plan (dorsal view, right digit). Bone resection includes the ulnar thirds of the distal part of the proximal phalanx, middle phalanx, and terminal phalanx and resection and fusion of the distal interphalangeal joint. Internal fixation can be performed using a K-wire and/or interosseous wire. Soft tissue is excised on the ulnar side, including the ulnar neurovascular pedicle and distal third of the nail bed, and closed with interdigitating palmar and dorsal flaps. The ulnar proximal interphalangeal joint collateral ligament, which is detached from the excised bone, is reinserted through drill holes. Six months postoperatively, the radial side is debulked, preserving the radial neurovascular pedicle. **B**, The postoperative appearance.

sive soft tissue reductions include excision of one digital nerve, as described by Tsuge,⁴⁹ Fujita et al,⁶³ Ogino,¹¹ and Bertelli et al.⁵⁰ The sensory results of this excision merit further study. Tailoring the nail plate is a component of most of the previously described procedures and contributes to normalizing the appearance of the fingertip. No technique is perfect, and most patients require multiple operations in a staged fashion.⁴⁸

POSTOPERATIVE CARE

It is possible to increase postoperative comfort by giving a digital or distal regional bloc at the end of surgery (for example, the median nerve or the ulnar nerve at the wrist) with a long-acting agent such as bupivacaine. The dressing should be sufficiently well padded to protect against pressure points, but it should not cause postoperative compression. This is particularly important when a bloc has been used. Surgeons must anticipate postoperative swelling, which may be substantial if the total tourniquet time is longer than 2 hours or if the surgery is extensive.

Patients who undergo soft tissue surgery alone, especially young children, often benefit from 7 to 10 days of immobilization, which provides comfort and incision protection. Osteotomies and epiphysiodeses necessitate at least 3 weeks of immobilization, depending on the presence of internal fixation and the stability of the technique used for osteosynthesis. Further, full-time or night splinting may be required to protect or adjust the position. Ligament reconstruction or repair should be immobilized for 4 to 6 weeks (4 weeks for patients 2 years of age and younger and 6 weeks for older patients). Subsequently, 6 weeks to 2 months of night splinting is recommended, and sports or other activities that may place strain on the repair are prohibited for this time. Patients who are not stiff and have an increased risk of instability should wear a splint dur-

ing the day and remove it only for scar massage and exercises. After this period, further splinting for up to 1 year is recommended during sports and activities that may place stress on the repair, but patients who have stiffness should not use a splint.

For smaller children or patients with compliance difficulties, we place a well-padded plaster sugar-tong splint. Before the splint is placed, a small amount of Mastisol adhesive or tincture of benzoin is applied to the nonoperated skin of the hand and forearm to minimize motion of the dressing. The plaster strips are applied on the adequately dressed, well-padded limb, beginning on the palmar aspect of the digits, continuing proximally to the elbow (flexed to 90 degrees), and around the distal arm to the dorsal surface, where they continue distally along the forearm to the wrist (Fig. 47-19).



Fig. 47-19 A left upper limb sugar-tong plaster splint seen from the ulnar side, with elbow flexed to 90 degrees.



Fig. 47-20 An elevation technique using a nonelastic cloth tube sleeve.

Regardless of the dressing, postoperative elevation of the limb is highly recommended. We follow the rule “hand higher than elbow higher than shoulder” and use a suspension technique in which a nonelastic cloth tube sleeve is attached around the chest (Fig. 47-20). Its distal end can then be attached to something solid at the desired height. The length of the sleeve between the tips of the fingers and the site of attachment should not be long enough to encircle the neck of the patient. Elevation is easily obtained using this technique and is continued for 7 to 10 days for soft tissue surgery patients and for up to 2 weeks or more for patients with more extensive procedures or pronounced edema. Subsequently, the end of the sleeve can be attached over the opposite shoulder to the back portion of the attachment around the chest, elevating the hand to shoulder level.

As soon as the initial dressing is removed, the patient begins occupational therapy, including splinting, scar massage, the use of topical sheet silicone and/or compressive gloves for the scars (see Chapter 48), and exercises. In my experience, and as others have reported, patients with macrodactyly can develop hypertrophic scars.⁹ Patients are followed until the scars have matured, which may take 1 to 2 years or more. Follow-up continues in the outpatient clinic until the end of growth.

TREATMENT OUTCOMES AND COMPLICATIONS

Despite numerous techniques described in the literature, surgical management of this malformation is difficult, with less than satisfactory results in many cases. The large number of techniques in the literature are proof of this problem. Most surgeons have limited experience with this malformation, because it occurs infrequently, further compounding the problem. Soft tissue debulking techniques alone do not control the increase in bony width and length but may be sufficient for more subtle cases. In more severe forms, techniques that reduce length frequently involve loss of a joint, but most do not correct excess width. Procedures that also address width risk the development of joint stiffness. An epiphysiodesis halts longitudinal growth but does not decrease width. The best outcomes are achieved with combination techniques that treat all aspects. Several operations are frequently required to obtain an acceptable result.⁴⁸

In addition to difficulties in obtaining satisfactory results, complications can occur. These include vascular compromise of the digit; skin flap necrosis; hypesthesia; areas of anesthesia; angulation deformities; malunion or, more rarely, nonunion of epiphysiodeses or osteotomies; limitation of motion or joint instability; hypertrophic scars; and complications related to the dressings and anesthesia, common to all hand surgery.

FUTURE DIAGNOSTIC AND TREATMENT MODALITIES

Further work in molecular biology³⁸ and genetics, such as continued research in somatic mutations,⁴¹ may lead to a better understanding of this heterogeneous condition and provide new treatment modalities.

In the meantime, perhaps more experience with sequential combination techniques such as the operation of Bertelli et al^{50,58} in younger patients, followed by epiphysiodeses and/or closing wedge osteotomies, may improve treatment of these challenging cases.

KEY POINTS

- Careful attention to signs of nerve compression is essential for timely investigation and surgical treatment as indicated.
- Distinguishing between static versus progressive forms of macrodactyly is critical. Sequential comparative bilateral AP radiographs of the hands allow measurements and calculations of percentage differences in the length of various bones (or entire digits as long as no angulation deformity is present). These radiographs are usually obtained at 1-year intervals or longer, unless a clinical concern arises. Progressive forms predictably require a greater number of interventions.
- Because of the complexity in treating macrodactyly, a well-integrated interdisciplinary approach is often useful. Pediatricians and/or medical geneticists can screen for syndromic cases and refer patients to other specialists or multidisciplinary clinics. Occupational therapists and/or physiotherapists can evaluate function, follow for deterioration, and help to plan treatment. Psychologists and/or social workers can assess a patient and family's ability to cope with the malformation and multiple proposed interventions, and support the patient during various stages of treatment and life's milestones, such as starting day care or grade school or entering high school.

For Patients Seen Soon After Birth

- Whether the operation described by Bertelli et al⁵⁰ would provide satisfactory results in a larger number of patients and/or in younger patients is not known, but the technique seems promising. The longitudinal resection should be performed on the side of the digit that requires less collateral ligament stability.
- Epiphysiodesis with or without a closing wedge osteotomy should be carried out to arrest longitudinal growth when the digit reaches adult length.
- Other operations may be required before or after the reduction procedures (for example, carpal tunnel release or isolated closing wedge osteotomies).
- Amputation of a digit may be a reasonable alternative in some cases and should be discussed with the parents as a possible outcome early in the child's care.

For Patients Seen at an Older Age

- The operation described by Bertelli et al⁵⁰ may be considered for patients who are not too old. (The patient described was 8 years of age.)
- Epiphysiodesis should be performed as described previously in the chapter if the patient's digit is equal in length to that of an adult.
- One of the combination techniques can be used in older patients.
- Amputation of a digit may be a viable option.

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48

Congenital Hand Anomalies: Undergrowth

E. Patricia Egerszegi



Although *undergrowth* is traditionally the fifth separate class under the International Federation of Societies for Surgery of the Hand (IFSSH) classification system for congenital hand anomalies,¹ hypoplasia of varying parts of the upper limb is one of the most common clinical features of most of the classes of hand malformations,^{2,3} with the exception of most cases of overgrowth. For this reason, the following malformations are discussed in other chapters in this book:

- Transverse arrest of development, the most severe forms of which are difficult, if not impossible, to differentiate from some forms of undergrowth (Chapters 42 and 51)
- Longitudinal arrest of development, including thumb hypoplasia and central ray deficiencies (Chapters 42 and 43)
- Clinodactyly (Chapter 44)
- Hypoplasia of duplicated elements, including hyperphalangism (Chapter 46)
- Hypoplasia found in constriction band syndrome (Chapters 44 and 49)
- Hypoplasia can also be associated with some giant congenital nevi⁴ (Group II: failure of differentiation of parts—congenital tumorous conditions; however, this subclass is not included in older versions of the Swanson classification system)⁴

Undergrowth can involve the whole limb, the whole hand, or segments thereof, and involves any, several, or all of the structures of the limb. It can be unilateral, bilateral, or involve all four limbs, whereas the distribution pattern can be specific for certain subtypes. It can be isolated or part of a syndrome (Table 48-1 and Box 48-1). In its mildest form, undergrowth requires no treatment; however, in its more severe forms, treatment can be very challenging.

Table 48-1 Associations, Syndromes, or Diseases of Brachydactyly*

Class	Syndromes ⁵⁻¹⁰	Class	Syndromes ⁵⁻¹⁰
I Generalized	Achondroplasia Diastrophic dwarfism Dyschondrosteosis Ellis-van Creveld syndrome Geleophysic dysplasia ^{11,12} Hereditary multiple exostosis Hypochondroplasia Metaphyseal dysostosis Mucopolysaccharidosis (gargoylism) Multiple epiphyseal dysplasia Ollier disease Orofaciodigital syndrome Peters Plus syndrome ¹³ Peripheral dysostosis Smith-Magenis syndrome ^{14,15} Spondyloepiphyseal dysplasia Spondylometaphyseal dysplasia Weill-Marchesani syndrome	IV Type D brachydactyly (stub thumb brachydactyly)	Robinow syndrome Rubinstein-Taybi syndrome Tabatznik syndrome (heart-hand syndrome II) Wiedemann syndrome ²⁴
II Underdeveloped distal phalanges	Cleidocranial dysostosis Coffin-Siris syndrome Fanconi anemia Fetal hydantoin syndrome ¹⁶⁻¹⁸ Gorlin-Chaudhry-Moss syndrome ¹⁹ Keutel syndrome Killian/Teschler-Nicola syndrome (or Pallister-Killian, or Pallister mosaic syndrome, or tetrasomy 12p) ^{20,21} Kirner's deformity Larsen syndrome Nail dysplasia/brachydactyly/distal phalanx syndrome ²² Pallister-Hall syndrome (with syndactyly) ²³ Progeria Pudiger syndrome Pyknodysostosis Sorsby syndrome	V Brachydactyly with polydactyly	Ellis-van Creveld syndrome Orofaciodigital syndrome
III Type A3 brachydactyly (short middle phalanx of little finger)	Coffin-Siris syndrome Cornelia de Lange syndrome Down syndrome Noonan syndrome Oculodentodigital syndrome Orofaciodigital syndrome I and II Otopalatodigital syndrome Poly-X syndrome Rubinstein-Taybi syndrome Russell-Silver syndrome TAR syndrome	VI Brachydactyly with syndactyly (sybrachydac- tyly)	Aarskog syndrome ^{25,26} Aglossia-adactylia Ankyloglossia superior Apert syndrome Cleft palate and lateral synechia Cleft lip Clubfoot ²⁷ Congenital cataract Cornelia de Lange syndrome Ear deformity Foot reduction abnormalities Glossopalatine ankylosis Hanhart syndrome Moebius syndrome Morning glory syndrome Poland syndrome (or sequence) Renal anomalies Submucous cleft palate ²⁷ Summit syndrome ²⁸ Undescended testis: ipsilateral ²⁷
		VII Cone-shaped epiphysis	Acrodysostosis Jackson-Weiss syndrome ²⁹ Langer-Giedion syndrome Myrhe-LAPS syndrome ³⁰ Trichorhinophalangeal syndrome
		VIII Miscellaneous	Bloom syndrome Cleft hand Du Pan syndrome Hand-foot-uterus syndrome Holt-Oram syndrome Osteochondrosis dissecans ³¹ Symphalangism Treacher Collins syndrome

*List includes sybrachydactyly but excludes brachymetacarpia.

LAPS, Laryngotracheal stenosis, arthropathy, prognathism and short stature; TAR, thrombocytopenia-absent radius.

The definition of some of the terms commonly encountered in descriptions of undergrowth of the upper limb^{6,10,32-34} can be found in Box 48-2.

Box 48-1 Some Syndromes or Diseases Associated With Brachymetacarpia* (Including the First Metacarpal)

Acrodysostosis	Cretinism	Multiple exostoses syndrome
Aicardi syndrome	Cri du chat syndrome (deletion 5p syndrome)	Orodigitofacial syndrome
Albright hereditary osteodystrophy	Deletion 4q syndrome	Otopalatodigital syndrome, type I
Banki syndrome	Deletion 18q syndrome	Otopalatodigital syndrome, type II
Basal cell nevus syndrome	Diastrophic dysplasia	Pallister-Hall syndrome (Hall syndrome)
Biemond syndrome	Duplication 9p syndrome	Poland syndrome (or sequence)
Bilginturan brachydactyly	Duplication 10q syndrome	Progressive myositis ossificans
Bixler syndrome	Duplication of uterus	Pseudohypoparathyroidism
Brachydactyly A1	Dwarfism	Pseudopseudohypoparathyroidism
Brachydactyly C	Dyggve-Melchior-Clausen syndrome	Pyruvate kinase deficiency
Brachydactyly E	Ectodermal dysplasia (Bork) ^{35,36}	Robinow syndrome
Campomelic dysplasia	Fanconi anemia	Ruvalcaba syndrome
Catel-Manzke syndrome	Fetal alcohol syndrome	Saethre-Chotzen syndrome
Chondrodysplasia punctata, X-linked dominant type	Gorlin syndrome	Schinzel-Giedion syndrome
Cockayne syndrome	Gorman syndrome	Silver syndrome
Coffin-Siris syndrome	Grebe syndrome	Sybert syndrome
Cohen syndrome	Holt-Oram syndrome	Tabatznik syndrome (heart-hand syndrome II)
Congenital glaucoma	Kabuki syndrome	Taybi-Linder syndrome
Congenital hemidysplasia with ichthyosiform erythroderma and limb defects (CHILD) syndrome	Klinefelter syndrome	Tibial aplasia—ectrodactyly syndrome
Conradis disease	Larsen syndrome	Trichorhinophalangeal syndrome, type I
Cornelia de Lange syndrome (Brachmann—de Lange syndrome, de Lange syndrome)	Leri pleonosteosis	Triploid syndrome
	Leri-Weill dyschondrosteosis	Tuomaala-Haapanen syndrome
	Levy-Hollister syndrome	Turner syndrome (XO, 45X)
	Mseleni joint disease	Ulnar-mammary syndrome
	Multiple epiphyseal dysplasia	Wiedemann syndrome

*Including features described by Gupta and Scheker,⁷ McCombe and Kay,⁸ Jones,³⁷ Jones,³⁸ and Wood.³⁹

Box 48-2 Commonly Used Terms for Describing Upper Limb Undergrowth

adactyly Absence of digits	monodactyly Presence, usually congenital, of only one digit on the hand or foot
brachydactyly Short digit	oligodactyly Subnormal number of fingers or toes (often used to be synonymous with atypical cleft hand and symbrachydactyly*)
brachysyndactyly Short and fused digits	peromelia Absence (or deformity) of the terminal part of a limb
brachymetacarpia Short metacarpal	symbrachydactyly* Previously known as atypical cleft hand (see brachysyndactyly)
brachyphalangia Short phalanx	syndactyly Fusion of digits
brachybasophalangia Short proximal phalanx	
brachymesophalangia Short middle phalanx	
brachytelephalangia Short distal phalanx	
clinodactyly Permanent radial or ulnar deviation of one or more fingers	
ectrodactyly Absence of all or part of a digit (usually used for group I—Failure of formation of parts—longitudinal deficiencies—central); occasionally used for transverse deficiencies)	

*Some authors still differentiate between *symbrachydactyly*, defined as a central loss with hypoplastic digits (and classify it under group I: Failure of formation—longitudinal arrest—central ray deficiency [cleft hand—atypical type]), and *brachysyndactyly*, defined as central syndactyly with short digits (and classify it under group V: Undergrowth—phalangeal [digit]),⁴⁰ rather than consider brachysyndactyly as a less severe form of symbrachydactyly, as done by Yamauchi and Tanabu¹⁰ and in this chapter.

CLASSIFICATION

The IFSSH¹ subclassifies undergrowth first into the area involved:

- 1 = Whole limb
- 2 = Forearm and hand
- 3 = Hand alone
- 4 = Metacarpal
- 5 = Digit

The IFSSH then subclassifies certain categories (Table 48-2). Although no classification system neatly organizes all of the subtypes, the two main categories involving the hand, *brachydactyly* (including brachymetacarpia) and *brachysyndactyly* (now known as *symbrachydactyly*), have each been subclassified by various authors.

Although the first radiographs of brachydactyly were published in 1896⁴¹ and the term coined the same year,⁴² it was not until 1932 that McArthur and McCullough⁴³ first classified the anomaly into two groups: present at birth and evident with growth. One of the first commonly used classification systems for brachydactyly was published by Bell in 1951.⁴⁴ This classification system has since been modified and eponyms added^{7,9,44,45} (Table 48-3), and is the most useful classification system historically from a genetic point of view.

Table 48-2 IFSSH Classification System for Undergrowth

IFSSH System ¹ With Modifications ⁴⁶	Author's Suggested Modifications to the Classification of Undergrowth
	Thoracic muscle $\pm$ chest wall underdevelopment (isolated) ^{47,48}
Whole limb	Whole limb
Forearm and hand	Forearm and hand
Hand alone	Hand alone
Entire	Entire
Partial	Partial
Metacarpal	Metacarpal
Brachymetacarpia	Brachymetacarpia: bilateral or unilateral
Fifth ray	Fifth ray
Other brachymetacarpia	Other isolated ray
Other	Multiple rays
Digit	Digit: most bilateral
Brachysyndactyly	Brachydactyly
With absence of thorax muscles (Poland syndrome)	Brachymesophalangia
Without absence of thorax muscles	Brachytelephalangia
Brachydactyly	Brachybasophalangia
Brachymesophalangia	Symbrachydactyly: most unilateral
Defect of two or more phalanges	With thoracic muscle absence $\pm$ chest wall underdevelopment (Poland syndrome)
Defect of proximal or distal phalanx	Without thoracic muscle absence
Other	

Table 48-3 Modified Bell Classification System for Brachydactyly

Classes	Particular Features
Type A	
A1: Farabee brachydactyly	Short middle phalanges in all digits, when severe, absent, or fused to distal phalanges; variable grades of severity exist; thumb and big toe proximal phalanges are also involved
A2: Mohr-Wriedt brachydactyly (brachymesophalangy II)	Short middle phalanges of index fingers (which can be delta shaped) and second toes; radial deviation of the index fingers; type A2 is the rarest form of brachydactyly
A3: Bauer brachydactyly-clinodactyly (brachymesophalangy V)	Short middle phalanges of little fingers (which can be delta shaped, causing classic little finger clinodactyly with radial deviation [NOTE: In Gupta et al, ⁷ incorrectly written as ulnar deviation]); transmission is autosomal dominant ⁴⁴ ; more rare in white and black populations (~1%) and very common in Japanese (~21%) ⁴⁹
A4: Temtamy brachydactyly (brachymesophalangy II and V)	Short middle phalanges of index and little fingers; depending on the author, ring fingers (with associated radial deviation) and even middle fingers can be involved; thumbs can present duplication of the distal phalanges; feet have short middle phalanges of all toes and other deformities (such as talipes calcaneovalgus); short stature may be present; autosomal dominant transmission
A5: Bass brachydactyly	Absence of middle phalanges and hypoplastic nails in all four limbs or only upper limbs; may have distal phalangeal hypoplasia or thumb distal phalangeal duplication; autosomal dominant transmission
Type B: Mackinder brachydactyly	Short middle phalanges, absent or rudimentary distal phalanges; nails of index finger through little finger often absent; distal phalanges of thumbs and big toes, when involved, are flat or duplicated; toes 2 through 5 are often involved but less so than fingers; can have symphalangism or syndactyly (which would then be symbrachydactyly ⁷)
Type C: Drinkwater brachydactyly	Short index finger and middle finger middle phalanges; delta middle phalanges of little fingers; hypersegmentation of the index finger and middle finger proximal phalanges; ulnar deviation of the index fingers; normal ring fingers; occasional shortening of metacarpals or symphalangism
Type D: Breitenbecher brachydactyly (stub thumb brachymegalo-dactylysm)	Also known as “Potter’s thumb” or “murderer’s thumb”; short, wide thumb distal phalanges with disproportion between base of distal phalanges and head of proximal phalanges (the base is wider); seen in Rubinstein-Taybi and Tabatznik (heart-hand syndrome II) syndromes and resembles thumb involvement in type B brachydactyly
Type E: Bell brachydactyly (brachymetacarpia-metatarsia)	Short metacarpals (one or more) with or without metatarsals; normal phalanges; often associated with short stature, can have hyperlaxity; often a feature of other types of brachydactyly in addition to several syndromes; when part of Albright’s hereditary osteodystrophy, it is X-linked; in Turner syndrome, it is associated with the XO chromosomal anomaly; autosomal dominant with variable expression in its isolated form
E1 ⁵⁰	Involves fourth metacarpals and metatarsals only
E2 ⁵⁰	Variable metacarpal involvement with some phalangeal shortening (second and fifth metacarpals, index and middle finger distal phalanges, index and little finger middle phalanges)
Other types	
Combined B and E: Pitt-Williams brachydactyly	Ulnar distal phalangeal shortening with involvement of one or more metacarpals; not associated with short stature
Sugarman brachydactyly	Proximal phalangeal shortening with symphalangism; duplication of first toes

On the other hand, *brachysyndactyly* (syndactyly), which was first described by Poland in 1841,⁵¹ was not classified until 1921 into those cases associated with a pectoral muscle defect (*Poland syndrome*) and those without.⁵² Blauth and Gekeler⁵³ later postulated that the condition had a spectrum of severity and classified the malformation into four types.^{10,32,53}

1. *Short finger*: Syndactyly and anywhere from mild brachymesophalangism to complete middle phalanx aplasia of one or several digits, with or without stiffness and/or syndactyly, excluding the thumb—the most common form (33% to 59%)³²
2. *Oligodactyly, cleft hand* type (previously known as *atypical cleft hand*, a term no longer in use): Absence of one or several central digits (11% to 27%)³²
3. *Monodactyly*: Presence of the thumb and replacement of the remaining digits with vestigial nail-bearing nubbins—the rarest form (11% to 17%)³²
4. *Peromelia*: Adactyly and small bumps, which may or may not bear vestigial nails—the second most common form (20% to 23%)³²

Yamauchi and Tanabu¹⁰ subsequently expanded on this idea by developing what is presently the most useful classification system for syndactyly. This system classifies the anomaly according to the specific anatomic features and level of bony absence, thus facilitating treatment planning¹⁰ (Table 48-4 and Fig. 48-1).

Table 48-4 Yamauchi and Tanabu's Classification System for Syndactyly

Classes (percentage in this series)	Particular Features*
Unilateral syndactyly (98%)	Least severe forms have cutaneous syndactyly; all sporadic
Triphalangeal type (16%)	Shortness of some phalanges (usually severe shortening of middle phalanx, moderate shortening of central three distal phalanges, mild in other phalanges); mild shortness of metacarpals; absence of pectoral muscles in 82%
Diphalangeal type (15%)	Absence of one phalanx (usually middle phalanx) in one or more digits (usually central three digits); in those digits where present, this phalanx is severely shortened; other phalanges and metacarpals shorter than in previous type; absence of pectoral muscles in 56%
Monophalangeal type (10%)	Only one phalanx present in one or more digits (usually middle finger, followed by ring and index fingers); severely shortened proximal phalanges of central three digits; shortened distal and moderately shortened proximal thumb phalanges; metacarpals shorter than in previous type. Absence of pectoral muscles in 27%.
Aphalangeal type (24%)	All phalanges absent in one or more digits (usually central three digits); severe metacarpal shortening in central three digits and moderate in border digits; moderate shortening of forearm bones in over half the cases; decreased presence of syndactyly; absence of pectoral muscles in 20%
Ametacarpia type (15%)	One (thumb) or no digits present; severely shortened or absent metacarpals; moderate shortening of forearm bones in most cases; absence of syndactyly; similar to transverse arrest of development; no absence of pectoral muscles in this series
Acarpia type (10%)	Partial or complete absence of carpal bones with partial or complete fusion between those remaining, when present; marked shortening of forearm bones in most cases; absence of pectoral muscles in 18%
Forearm amputation type (8%)	Marked shortening of forearm bones; no absence of pectoral muscles in this series
Bilateral syndactyly (2%) (as high as 7% in other series)	Must differentiate from constriction band syndrome and cleft hand; no absence of pectoral muscles in this series

*Definitions of shortening as a percentage of the normal side: severe $\geq 65\%$; moderate 66% to 80%; mild 81% to 95%.



Fig. 48-1 Yamauchi and Tanabu's classification system for symbrachydactyly (Blauth and Gekeler's classification indicated in parentheses). **A** and **B**, Posteroanterior radiograph and clinical photograph of triphalangeal symbrachydactyly (*short-finger type*) of the right hand. The malformation is limited to mild skeletal shortening affecting the middle phalanges, similar to that in brachydactyly. There is also the presence of frequently encountered clinodactyly of the index through little fingers. **C** and **D**, Posteroanterior radiograph and clinical photograph of diphalangeal symbrachydactyly (*short-finger type*) of the left hand. **E** and **F**, Posteroanterior radiograph and clinical photograph of monophalangeal symbrachydactyly (*short-finger type*) of the left hand. The clinically palpable proximal phalanx of the long finger is not yet calcified and thus is not visible. **G** and **H**, Posteroanterior radiograph and clinical photograph of aphenalangia symbrachydactyly (*oligodactyly*) of the left hand. Marked clinodactyly of the index finger is evident. **I** and **J**, Posteroanterior radiograph and clinical photograph of ametacarpia symbrachydactyly (*oligodactyly*) of the left hand. Typical clinodactyly of the little finger in radial deviation and central digital nubbins is present.

Continued

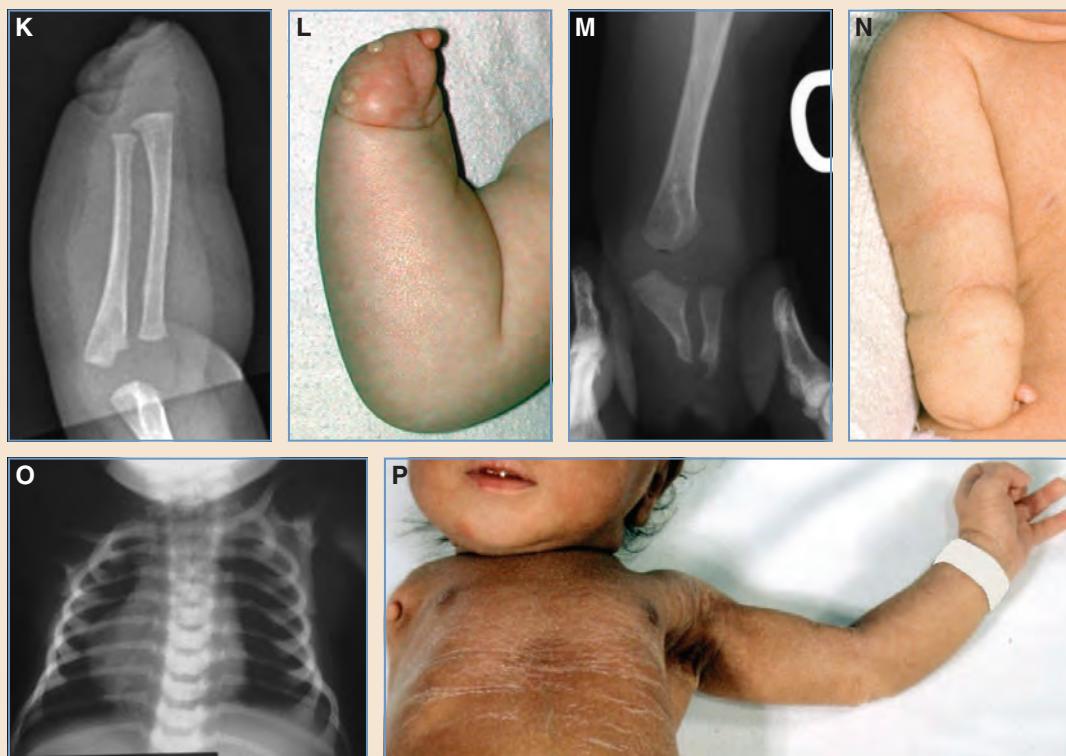


Fig. 48-1, cont'd **K** and **L**, Anteroposterior radiograph and clinical photograph of acarpia symbrachydactyly (*peromelia*) of the right forearm and hand. None of the carpal bones are visible, because they have not yet ossified. **M** and **N**, Anteroposterior radiograph and clinical photograph of forearm amputation symbrachydactyly (*peromelia*) of the right upper limb. **O**, Anteroposterior radiograph with complete absence of the upper limb and clavicle. **P**, Complete absence of the right upper limb with a vestigial nubbin at the shoulder and a normal left upper limb. **NOTE:** Although this form is not part of the original Yamauchi and Tanabu classification system, it is hereby suggested that it be included and has been added for completeness.

In the past, *brachysyndactyly*, which means short webbed fingers, was considered to be different from atypical cleft hand⁵⁴ and transverse deficiencies.^{1,55} Blauth and Gekeler⁵³ suggested that they were all variants of the same teratologic entity and named them *symbrachydactyly* (the term that will be used from here on). They have in common central ray reduction anomalies with developmental arrest of the second, third, and fourth digits.⁵⁵ The term *atypical cleft hand* (formerly synonymous with what is now *oligodactylic symbrachydactyly*)^{32,56} in the Blauth and Gekeler classification system,⁵³ was abandoned by the Congenital Hand Committee of the IFSSH in 1992.^{32,57} The terms *monodactylic* and *peromelic symbrachydactyly* (in the Blauth and Gekeler classification system⁵³) have likewise replaced certain terminal transverse arrests.³² *Symbrachydactyly* should therefore replace *brachysyndactyly* in the IFSSH classification system; the term should probably be dropped to avoid confusion, or at most should refer only to what used to be known as *short-finger brachysyndactyly* in the Blauth and Gekeler classification system⁵³ (see Table 48-2).

PATIENT CHARACTERISTICS

Because various types of undergrowth are a feature of many other congenital hand malformations, the incidence of undergrowth is unknown in its most generalized form. Some investigators have nevertheless reviewed their experience in congenital hand anomalies to try to establish the frequency of various malformations. In Flatt's series⁵⁸ of 2758 cases of hand malformations, the incidence of brachydactyly was 5.2% (143 cases), making it the sixth most frequent of all of the congenital anomalies, and the incidence of Poland syndrome was 2.2% (60 cases). Because amputations (186 cases [6.8%]) and whole-hand hypoplasia (27 cases [1.0%]) were in separate classes, it is possible that some of these were in fact cases of undergrowth. Flatt⁵⁸ also referred to the Yokohama series, in which brachydactyly was the third most common deformity (19 [7.0%] of 256 cases), and brachysyndactyly the tenth most common (10 [4.0%] of 256 cases). In this series, amputation (16 [6.0%] of 256 cases) and microdactyly (5 [2.0%] of 256 cases) were likewise classified separately, possibly including some cases of undergrowth. Cheng et al² noted a 4.3% incidence of undergrowth among 578 patients with upper limb congenital anomalies who were seen over a 10-year period at their center. Ogino et al⁵⁵ found the incidence of symbrachydactyly to be 4.5% (43 cases/955 hand anomalies) and brachydactyly 4.0% (38 cases/955 hands) in patients seen over a 16-year period.

Although these numbers give some idea of the proportion of hand malformations that these deformities occupy, they do not reflect the actual incidence in the general population. Because some congenital hand malformations are dealt with in the neonatal nursery, they may never be accounted for, such as narrow pedicled polydactyly. Other deformities are missed, such as mild brachydactyly, whereas others are never seen in consultation, such as mild clinodactyly or severe deformities with life-threatening associated conditions. In addition, variability among observers, variability by the same observer over time, and differences among various ethnic backgrounds² and areas of the world may also affect the total number of hand malformations registered, making precise statistics impossible.

Additional factors that confuse the picture include difficulties in classifying certain malformations such as symbrachydactyly, which are variously classified as: failure of formation—transverse arrest; failure of formation—longitudinal arrest—central ray deficiency (cleft hand)—atypical type; and undergrowth.⁴⁶ Cheng et al² underlined this problem when trying to classify the spectrum of transverse arrests, atypical cleft hand, and brachysyndactyly. Furthermore, modifications in the classification system itself have contributed to the confusion rather than clarified the picture. In the past, some authors have placed symbrachydactyly under failure of formation—longitudinal arrest—central ray deficiency (cleft hand)—atypical type, and brachysyndactyly under undergrowth—phalangeal (digit),⁴⁰ claiming that symbrachydactyly, brachysyndactyly, and brachysymphalangism should not be grouped together.⁵⁹ Others argue that brachysyndactyly, atypical cleft hand, and transverse deficiencies are all variants of the same teratologic entity and should all be called *symbrachydactyly*.⁵³ In this chapter, I think that brachysyndactyly, atypical cleft hand, and certain transverse deficiencies are variations of symbrachydactyly and should all be classified under group V: Undergrowth, in the IFSSH system.

There are few reports of actual incidences of certain congenital hand malformations in specific general populations.^{60,61} Castilla et al⁶⁰ evaluated a total of 599,109 live births in 60 South American hospitals participating in the Latin American Collaborative Study for Congenital Malformations. The incidence of symbrachydactyly with Poland syndrome was 0.00002% (12/599,109). Unfortunately, it is unclear whether all cases of symbrachydactyly without Poland syndrome are

accounted for in this paper (if so, its incidence would be 0.00003% [18/599,109]), and it is possible that mild cases of both Poland syndrome and isolated symbrachydactyly were missed.⁶⁰ In addition, several patients who would likely be included in this category today were possibly classified elsewhere. In the report by Freire Maia and Azevedo,⁶¹ a house-to-house survey in central Brazil revealed two cases of Poland syndrome in 58,761 births (0.00003%), whereas cases now considered symbrachydactyly are grouped variably in other categories and cannot be extracted from the data. Although these papers provide some idea of the frequency with which symbrachydactyly in Poland syndrome occurs, they do not refer to other forms of undergrowth, nor do they provide insight about the percentage of all congenital hand anomalies with this diagnosis.

In general, it can be said that brachydactyly, as classified by Bell and modified by subsequent investigators,^{7,9,44,45} refers mostly to bilateral deformities, some involving all four limbs, with various known or suspected inheritance patterns. Many of these are autosomal dominant, and several can be associated with various syndromes (see Table 48-1 and Box 48-1) (Fig. 48-2). The latter vary in inheritance patterns from autosomal dominant (such as Apert syndrome,⁶² which is best classified under group VII: Generalized skeletal abnormalities, but is included by some in group V: Undergrowth) to sporadic (such as Ollier disease).⁶³

Brachymetacarpia, which can be unilateral, bilateral, or can even involve one or both feet, is frequently associated with phalangeal hypoplasia⁶⁴ and thus brachydactyly. It is found in numerous diseases and syndromes^{7,8,37-39} (see Tables 48-1 and 48-3), some of which have an autosomal dominant pattern of heredity. It is of note that little-finger brachymetacarpia is more common (64%) in children with familial short stature compared with those with normal growth (21.7%).^{65,66} Symbrachydactyly (brachysyndactyly), on the other hand, is much more frequently unilateral and sporadic. It may or may not be associated with ipsilateral pectoral hypoplasia (that is, Poland syndrome, which varies in frequency from 22%¹⁰ to 40%⁶⁰ of cases of symbrachydactyly, depending on the study).

There are some forms of digital undergrowth that do not neatly fit into any of these categories and nevertheless qualify as undergrowth. There is little information regarding sex distribution in most of the references cited, with the exception of Castilla et al,⁶⁰ who noted a higher male prevalence for symbrachydactyly with or without Poland syndrome (60%).

Various anomalies have been found to be associated with undergrowth. Clinodactyly, symphalangism, syndactyly, and pectoral muscle hypoplasia (the latter two now considered features of symbrachydactyly),⁸ as well as polydactyly, cleft hand (the latter possibly referring to some

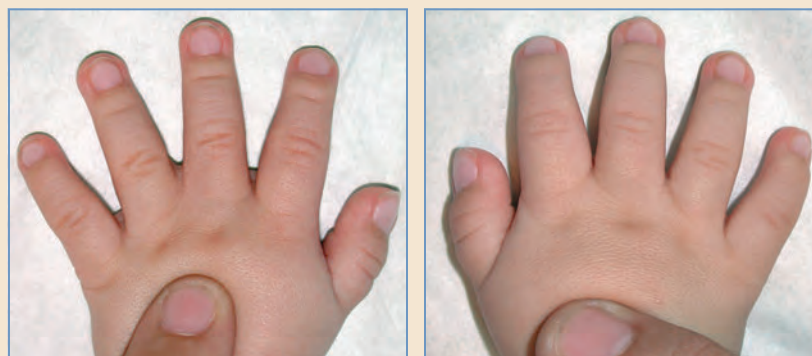


Fig. 48-2 Bilateral brachydactyly. The left and right hands of a patient with trisomy 9.

cases of “atypical” cleft hand, now considered a form of symbrachydactyly), Kirner deformity, and osteochondrosis dissecans of the finger,⁹ among others, can be associated with brachydactyly. Adactylia, peromelia, foot reduction anomalies, clubfoot, ear malformations, cleft lip or palate, Moebius syndrome, and ipsilateral renal hypoplasia or aplasia have been reportedly associated with symbrachydactyly,¹⁰ as have the following syndromes: aglossia-adactylia, ankyloglossia superior, glossopalatine ankylosis, Hanhart syndrome, cleft palate–lateral synechia, morning glory syndrome, and Poland syndrome, among others.³² It is important to note that the previously described associated anomalies and syndromes are subject to the difficulties caused by confusion over previous methods of classification of undergrowth. Hopefully there will be eventual reorganization, clarification, and worldwide adoption of a better classification system that will permit clearer associations between anomalies or syndromes and the various subclasses of undergrowth.

Hand Characteristics

Brachydactylies, as classified by Bell and previously mentioned, are frequently bilateral, and some also involve the feet. The bones most frequently affected are the middle phalanges, which are also the last to ossify.⁶ The index and little fingers are the most frequently implicated with this anomaly.^{6,64} After little-finger middle phalanx reduction, the next most frequent form of hypoplasia is that of the distal phalanx.⁶ These fingers may or may not be associated with nail hypoplasia or aplasia.⁶ There can be associated clinodactyly, symphalangism, or syndactyly⁸; when syndactyly is present, the case is likely a form of symbrachydactyly. Specific forms of brachydactyly can have characteristic involvement of other bones and/or rays in addition to those previously mentioned (see Tables 48-1 and 48-3).

As classified by Yamauchi and Tanabu,¹⁰ symbrachydactyly is most often unilateral, having been identified 95.8% of the time when they combined their own cases with those they refer to in the literature, although bilateral cases do occur (Fig. 48-3). Depending on the reference, unilateral involvement is either relatively equal in distribution between the left and right sides,⁶⁷ more often found on the left (60.6% to 75% of cases),^{10,53} or even more frequent on the right (73.3% if both symbrachydactyly and Poland syndrome are combined; 75% if only Poland

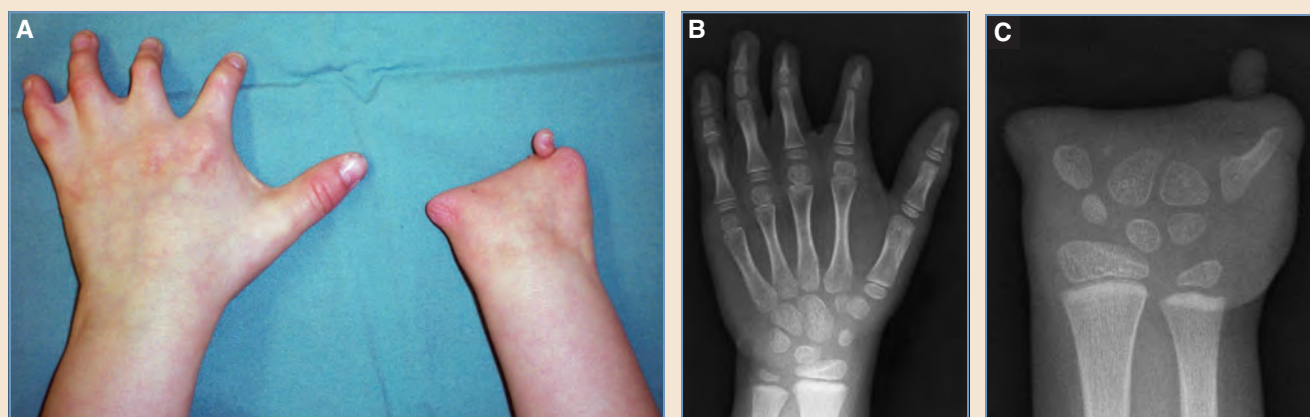


Fig. 48-3 Bilateral symbrachydactyly. **A**, Diphalangal type on the left and acarpia type on the right, as classified by Yamauchi and Tanabu. These would be classified as short finger type on the left and peromelia on the right in Blauth and Gekeler's classification system. **B**, Posteroanterior radiograph of the left hand. **C**, Posteroanterior radiograph of the right hand.

Table 48-5 Features Aiding in Differentiation Between Symbrachydactyly, Cleft Hand, and Amniotic Band Syndrome*

Features	Symbrachydactyly	Cleft Hand	Amniotic Band Syndrome
Heredity	Usually sporadic	Frequently hereditary (autosomal dominant)	Sporadic
Limbs involved	Usually one limb (frequently upper, but lower has been reported ⁶⁸)	One to four (frequently bilateral upper limbs, feet often involved)	One to four in any combination of upper and/or lower limbs
Proximal upper extremity	Forearm/arm hypoplasia; frequent pectorothoracic abnormality	Normal	Normal
Shape of defect	U	V	Frequently straight line (transverse or oblique)
Nubbins	Present in oligodactylic (aphalangia) type; frequent presence of fingernails; no edema	No	Occasional (associated with amniotic band mark); rare presence of fingernails except distal to amniotic band mark if distal phalanx is present; can have edema distal to amniotic band mark
Acrosyndactyly†	No	No	Occurs
First web syndactyly	Mild or none	Occurs	Rare (if so, acrosyndactyly)
Bony abnormalities	Middle, proximal, then distal phalanx, followed by metacarpal hypoplasia; no transverse bones; no synostosis (osseous syndactyly)	Distal to proximal involvement; transverse bones possible; possible synostosis (osseous syndactyly)	Distal to proximal involvement; “jumbled” bones possible; possible osseous acrosyndactyly
Direction of digital involvement with increasing severity	Radial to ulnar (last remaining single digit is the thumb); begins with index and middle fingers	Central to radial (last remaining digit is the little finger); begins with middle finger	None
Number of rays involved	Multiple	Few	Variable, but often involves the longest digits (index through little fingers)
Metacarpal length	Shorter than normal	Normal or longer than normal	Normal

*Including features described by Büchler,³² Kay and McCombe,⁵⁶ and Buck-Gramcko⁶⁹ with modifications.

†Syndactyly between the distal parts of the digits with persistence of a slit or sinus between the bases of the involved digits.

syndrome cases are included).⁶⁰ According to Yamauchi and Tanabu,¹⁰ the malformation spans a spectrum of severity ranging from simple reduction in length, without any bony absence, to loss of part of the forearm (Table 48-5). They stated that the order of bony hypoplasia, followed by absence, begins with the middle phalanx, progresses through the presence of only one phalanx (the proximal one), loss of the metacarpal,⁷⁰ loss of carpal bones, and finally severe shortening of the forearm.¹⁰ It is hereby suggested that complete absence of the limb with one or several small nubbins present at the shoulder be considered part of this progressive sequence (see Fig. 48-1, *O* and *P*). The middle finger ray is the first affected, followed by the index and ring finger rays, then the little finger, and finally the thumb ray.¹⁰ Vestigial elements of digits remain at the distal end of the limb in the form of nail elements and/or soft tissue “buds.”^{70,71} Castilla et al⁶⁰ mentioned that there may be nail hypoplasia of the index finger or thumb. Clinodactyly of the remaining digits is not uncommon^{32,60} (see Fig. 48-1, *G* and *H*), whereas joint stiffness (often interphalangeal) or instability (metacarpophalangeal or interphalangeal) can occur, particularly in the little finger in oligodactylic types.³² More proximally, in the forearm, there may be minor hypoplasia,³² which is progressively more pronounced with increasing severity of the malformation.¹⁰

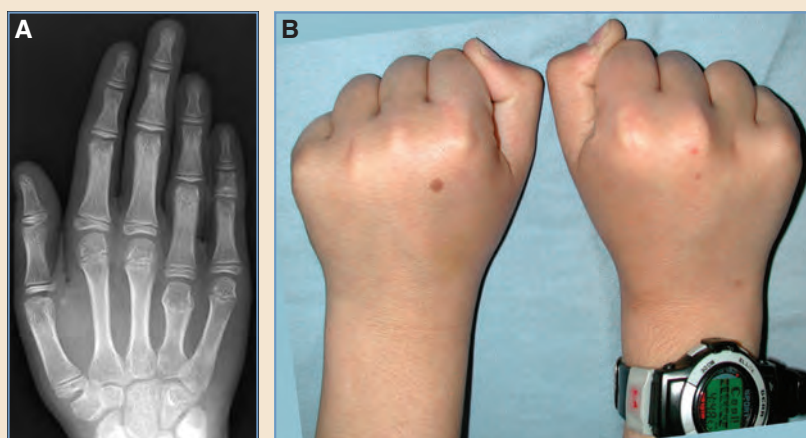


Fig. 48-4 Bilateral metacarpal hypoplasia most pronounced in the right fourth metacarpal, but also affecting the third metacarpal and the left hand to a lesser extent. **A**, Posteroanterior radiograph of the right hand. **B**, Clenched fists showing the marked right fourth brachymetacarpia.

The thoracic muscles (particularly the sternocostal head of the pectoralis major), the breast, and even elements of the chest wall may or may not be absent or underdeveloped.^{10,32,71} There have been cases reported of isolated partial absence of the pectoralis major muscle with associated breast asymmetry without hand involvement,⁴⁸ and even a case of isolated pectoralis muscle absence in a patient whose second cousin had full Poland syndrome.^{47,72}

Isolated brachymetacarpia tends to affect the ulnar two rays more so than the radial rays.^{6,9} Nevertheless, any single ray can be involved in isolation; the third through fifth rays together can be hypoplastic, as can all the metacarpals.³⁹ Other combinations of several metacarpals have also been described^{6,7,9} (Fig. 48-4). When isolated, isolated brachymetacarpia rarely has a significant functional impact.^{6,39} There is little information in the literature regarding the relative incidence of bilaterality versus unilaterality, and sidedness in unilaterality, for isolated brachymetacarpia.

ETIOLOGIC FACTORS AND PATHOLOGY

Many *bilateral* or *four-limb malformations* (that is, brachydactylies and brachymetacarpias) have been shown to have a genetic cause resulting from various mutations or deficiencies in cartilage-derived morphogenic protein, such as Grebe and Hunter-Thompson chondrodysplasias,⁸ or the bone morphogenic protein pathway.⁷³

Among unilateral deformities, such as most cases of symbrachydactyly, there seems to be a mesodermal abnormality that explains the persistence of ectodermal structures (that is, pulp, nails).⁸ The hypothesis is that the skeletal precursor is abnormal in differentiation or formation with secondary soft tissue deficiency.³² There is a theory that Poland syndrome, as well as Klippel-Feil anomaly, Moebius syndrome, isolated absence of the pectoralis major with breast hypoplasia, isolated transverse limb defects, and Sprengel anomaly, all result from early interruption of the vascular supply in the subclavian artery, vertebral arteries, and/or their branches.⁷⁴ Although there is some support for this, including impedance plethysmography results in eight patients with Poland syndrome, as reported by Bouvet et al,⁷⁵ consistent with hypoplasia of the subclavian artery on the affected side, most published cases have had no vascular imaging. There was even a case published in which the abstract states that there was no arterial anomaly, but it does

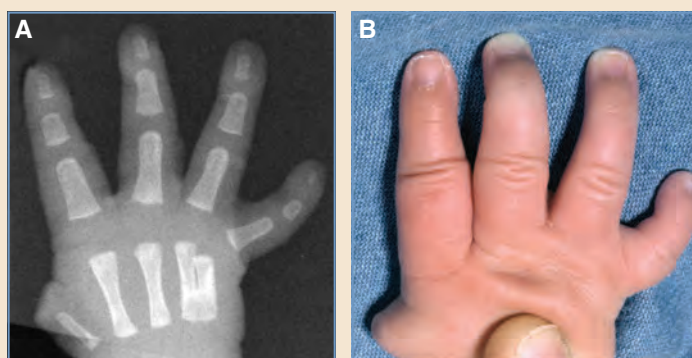


Fig. 48-5 Brachymetacarpia with associated fusion of the fifth metacarpal to the adjacent fourth metacarpal in a patient with thumb hypoplasia. **A**, Posteroanterior radiograph of the right hand. **B**, Right hand.

not specify how the arterial system was imaged, nor even refer to this point in the article text.⁷⁶ There have been published cases equivalent to Poland syndrome of the lower limb with unilateral gluteal hypoplasia and brachysyndactyly (sybrachydactyly)^{68,77} and cases of sybrachydactyly with involvement of both the hand and foot.^{78,79} There have even been reports of familial cases of upper limb Poland syndrome⁸⁰ and of gluteal involvement with phenotypic heterogeneity.⁸¹ These cases provide support for a genetic cause in certain patients with Poland syndrome.

Although sporadic cases may be multifactorial, a genetic error during mitosis generating a mosaic of somatic daughter cells that influence the development of the limb could be a factor.⁸⁰ The timing of this error might explain the phenotype and severity of the malformation.⁸⁰ The vascular anomaly, when present, might be the result of this process rather than its cause.

Anatomically, brachydactyly and brachymetacarpia, in their general forms, are limited to shortening and/or hypoplasia of the involved skeletal element, regardless of whether this includes the associated joint or joints, such as the fusion of two phalanges or symphalangism⁷⁻⁹ (see Table 48-4). Involved phalanges may have a delta abnormality with associated clinodactyly⁷⁻⁹; that is, in increasing order of severity, the abnormal bone varying from a trapezoidal bone, a longitudinal epiphyseal bracketed bone, a triangular bone (see Chapter 45) according to the subclass. Nails may be normal, hypoplastic, or absent according to the subtype⁷⁻⁹ (see Table 48-4). Hypersegmentation of the proximal phalanx is a feature of type C Drinkwater brachydactyly, in which there may also be deviation of the index finger in an ulnar direction.⁶ Brachymetacarpia may occasionally include fusion to the adjacent metacarpal⁶ (Fig. 48-5), a feature which can occur as a separate entity, a form of ulnar hypoplasia, or an anomaly common to several other hand malformations.⁹ Some forms, such as type B Mackinder brachydactyly, can have associated syndactyly (see Chapter 45 section on soft tissue abnormalities) and therefore should be classified as sybrachydactyly.⁷

In cases of sybrachydactyly, anatomic anomalies may be limited to mild skeletal shortening, similar to brachydactyly, in the triphalangeal type, affecting the middle phalanges (see Fig. 48-1, *A* and *B*), with or without flexion contractures at the interphalangeal joints, or stiffness, particularly at the distal interphalangeal joints.⁸² Even the thumb may have some signs of hypoplasia, and its metacarpophalangeal joint may also be stiff.⁸² Some degree of clinodactyly may be present with ulnar deviation of the index finger and radial deviation of the ring finger.⁸² In some cases, the radial two digits (excluding the thumb) may deviate ulnarly and the ulnar two may deviate radially (see Fig. 48-1, *A* and *B*), but there are exceptions (Fig. 48-6).

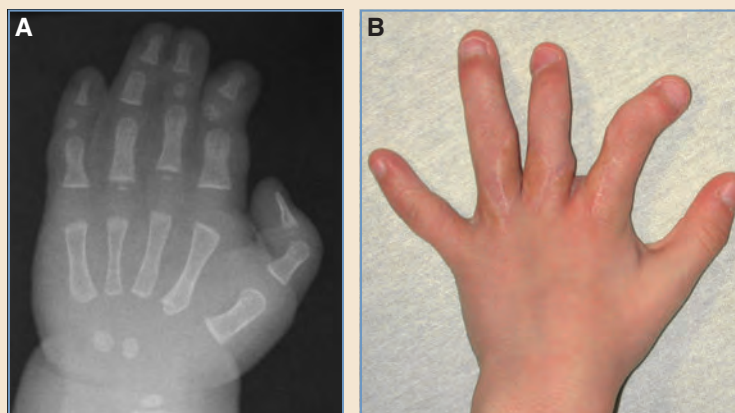


Fig. 48-6 Triphalangeal symbrachydactyly. **A**, Posteroanterior radiograph of the left hand showing clinodactyly with radial deviation of the index finger. **B**, Left hand after sequential syndactyly releases; the index finger would benefit from correction for clinodactyly.

As severity increases, the middle phalanges of the involved digits are absent (diphalangeal type) (see Fig. 48-1, *C* and *D*). The central digits are usually more severely affected than the border ones,¹⁰ although some authors note more involvement of the radial digits (excluding the thumb).^{32,69} The interphalangeal joints may be stiff or unstable.⁸² There is a certain degree of clinodactyly of the digits, particularly of the little finger.⁷¹ There is often syndactyly, particularly but not always⁶⁴ at the fourth web.⁷¹ As severity progresses, the affected digits contain a single phalanx (monophalangeal type) (see Fig. 48-1, *E* and *F*), and are replaced by soft tissue nubbins in the aphalangia type, while maintaining vestigial nail elements^{10,32} (see Fig. 48-1, *G* and *H*). The metacarpals become progressively more hypoplastic as the severity increases.^{10,71}

The digital nubbins can be attached to the extrinsic flexor and extensor tendons through an aponeurotic system. The flexors are often conjoined to various degrees⁸² and activated by a common muscle mass.^{71,82} The extensors, on the other hand, tend to remain separate, including the extensor indicis proprius and extensor digiti minimi tendons.⁷¹ Despite the presence of several vestigial digital nubbins, the little finger remains the best preserved of the digits excluding the thumb. It can have a hypoplastic or absent middle phalanx and a variably stable (unstable to stiff) single interphalangeal joint.^{71,82} The digit tends to deviate in a radial direction because of the pull of the extrinsic extensor (or extensors),^{32,71} and it may have little or no active flexion in some cases⁸² or instability in others.³² The thumb may have hypoplastic phalanges (more so at the proximal phalanx level) or an absent proximal phalanx,⁷¹ and may even have characteristics of thumb hypoplasia.^{71,82} The nail and pulp are smaller.⁷¹ When the distal phalanx is present, the interphalangeal joint can be unstable, but the basal joint may have adequate motion and stability.⁷¹

As severity increases, the little-finger phalanges are reduced to one and finally disappear, leaving a soft tissue nubbin in place of the finger. All of the metacarpals are hypoplastic, even that of the thumb, which then has an underdeveloped basal joint.⁷¹ The thumb proximal phalanx may be present or even absent,⁷¹ and the distal phalanx may be hypoplastic (see Fig. 48-14, *A* and *B*). The intervening joint is hypoplastic and often unstable. The extrinsic flexors of the absent fingers are conjoined into a mass, as are the extrinsic extensors, although the extensor indicis proprius and extensor digiti minimi may still be distinct.⁷¹ The flexors and extensors are fused together through an aponeurotic plane that produces variable amounts of motion or puckering of the

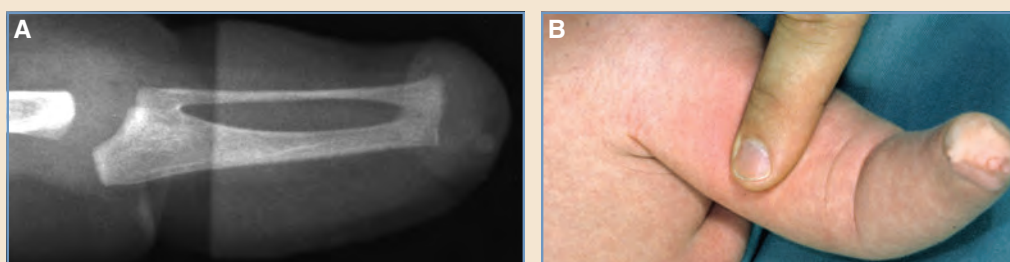


Fig. 48-7 Acarpia symbrachydactyly (peromelia). **A**, An anteroposterior radiograph of the left upper limb, in which there is synostosis between the radius and ulna. **B**, Left upper limb.

finger nubbins.⁷¹ The wrist tendons, particularly the extensors, tend to remain relatively normal.⁷¹ The greater the motion—that is, flexion and extension—of the nubbins or wrist, the more likely it is that the involved tendons have useful excursion.⁷¹

Finally, in the peromelic cases (ametacarpia [see Fig. 48-1, *I* and *J*], acarpia [see Fig. 48-1, *K* and *L*], and forearm amputation types [see Fig. 48-1, *M* and *N*]), there is transverse absence at or proximal to the metacarpal row. Synostosis can occur between the remaining hypoplastic metacarpals, but more frequently between the carpal bones,⁸² and even between the radius and ulna (Fig. 48-7). Wrist tendons may remain⁷¹ if some bony elements of a wrist are present. Vestigial digital tendons with minimal excursion may still be present.⁷¹ The most severe form of symbrachydactyly (rather than a subclass of failure of formation—transverse arrest) is one or several vestigial nubbins attached to the shoulder with complete absence of the humerus and all other elements distally (see Fig. 48-1, *O* and *P*).

In terms of innervation, in more severely affected hands, useful intrinsic function is absent and nerve abnormalities occur.⁷¹ The radial nerve remains, but the ulnar nerve, and with absence of the thumb, even the median nerve are hypoplastic or absent.⁷¹ Rarely, all major nerves in the hand are absent.⁷¹ In cases of undergrowth, vascularization can also be abnormal. The division of the common digital artery into the two proper digital arteries can be more distal than usual, particularly in symbrachydactyly.^{4,69} In rare cases of symbrachydactyly, one of the two proper digital arteries may even be absent.⁴

PATIENT ASSESSMENT

It is essential to examine the hands in detail. This includes determination of which rays are hypoplastic, to what extent, and at which level; the presence of thenar (or even hypothenar) hypoplasia; digital malposition, such as thumb adduction with or without supination⁸²; the presence of syndactyly, to what extent, and in which spaces; joint stability and mobility, both active and passive; and finally, hand function, including tendencies, compensations, and problems. An occupational therapy assessment is very helpful in evaluating the hands and can help in the development of an appropriate treatment plan.

Next, it is essential to examine the rest of the limb, pectoral girdle muscles, and thoracodorsal structures to check for limb length discrepancy or Poland syndrome (Fig. 48-8). Poland syndrome usually involves a pectoralis major muscle defect (usually absence of the sternocostal head, but possibly more extensive) and associated symbrachydactyly (that is, often index, middle, ring, and little finger brachydactyly, occasionally thumb brachydactyly, digital syndactyly, and hand

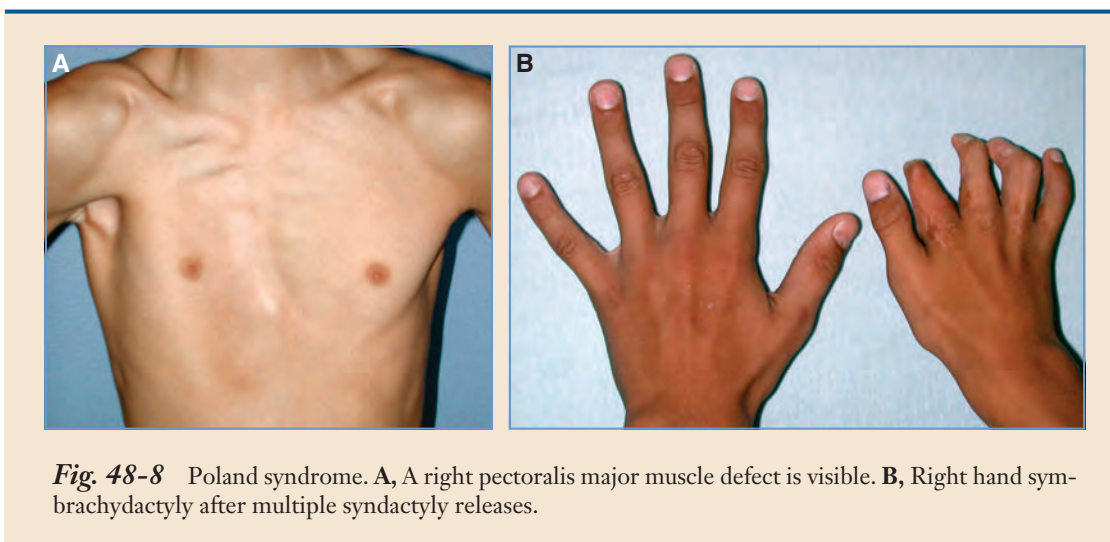


Fig. 48-8 Poland syndrome. **A**, A right pectoralis major muscle defect is visible. **B**, Right hand symbrachydactyly after multiple syndactyly releases.

hypoplasia).⁷² There may also be other pectorothoracic anomalies, such as breast hypoplasia,⁸³ nipple and/or rib hypoplasia,⁶⁰ absence of the pectoralis minor muscle,⁸⁴ latissimus dorsi muscle hypoplasia,⁸³ Sprengel deformity, scoliosis,¹⁰ or hypoplasia of the ipsilateral lower limb with absence of toes.⁸⁵

Because many associations, diseases, and syndromes (see Table 48-1 and Box 48-1) can accompany certain undergrowth deformities, it may be appropriate to refer the patient to a medical geneticist, other specialists, or an interdisciplinary team to evaluate, treat, and follow associated problems. Finally, it is helpful to identify the exact diagnosis for the hand anomaly, if possible, to help aid in counseling and management of the child and family. Such features as unilaterality/bilaterality, involvement of the lower limb (or limbs), and pectoral and/or pelvic girdle help differentiate between brachydactyly and symbrachydactyly. Nevertheless difficulties can arise occasionally in distinguishing between symbrachydactyly, cleft hand, and amniotic band syndrome. Several features can help differentiate between these malformations (see Table 48-5).

Comparative radiography of the normal (if present) and involved sides (including, if indicated, and in cases of symbrachydactyly, scanography to determine the presence of a limb length discrepancy) can help establish the diagnosis and determine the level of the anomaly and its severity. If radiography is performed very early, such as shortly after birth, calcification may not be sufficiently advanced, especially in the affected bones, to determine the full extent of the malformation or the true size of the affected bones (see Fig. 48-1, *E*). For this reason it is frequently preferable to wait until 6 to 8 months of age or even later before performing radiography, particularly when no surgery is planned at a young age. In cases in which a toe phalanx transfer is planned, anteroposterior radiography of the toes close to the time of surgery should be performed to choose the most appropriate toe phalanx in width and length for the procedure.⁸⁶ When the patient is very young, significant amounts of these bones are still cartilaginous; therefore portions are not yet visible. This must be taken into account when choosing a toe phalanx, because the actual bone is longer and wider than that measured. Similarly, it may be preferable to repeat earlier radiography or wait until just before surgery to obtain certain hand radiographs to better evaluate the malformation for surgery. When clinical signs warrant, medical imaging (such as a scoliosis series, CT scanning, or MRI) is useful to establish the extent of the malformation in the trunk. In addition, occasionally, psychological evaluation and follow-up support may be an essential component of the treatment plan for certain patients.

TREATMENT OPTIONS

There are many cases of mild undergrowth that require no treatment of any kind. Some mild cases of joint stiffness or flexion contracture can benefit from stretching exercises and/or splinting in occupational therapy. Patients with more severe undergrowth are best left alone in the following circumstances: when the risks or complexity of the possible surgical procedures outweigh the gains, when parental or patient expectations are greater than what can be realistically achieved, when surgery is not desired by the family or patient, in an adult patient who functions well and has become accustomed to the deformity, or when serious health conditions dictate. In most cases, surgery must be tailored to the individual patient's needs and wishes, the deformity, and functional problems.

PREOPERATIVE PLANNING

Surgical treatment can be subdivided into four general categories: web-deepening procedures (most often syndactyly releases), lengthening techniques, joint stabilization operations (usually fusions), and other corrective procedures including tendon transfers or even amputations.

Surgical Plan

The actual choice of procedure depends, at least in part, on the following: the age at presentation; presence of bilaterality; involvement of the feet; severity, number, and size of the digits; presence and degree of clinodactyly; active range of motion at each joint; and presence of joint instability.

When a single digit is shorter, especially the little finger, although no treatment is generally indicated, power grip strength may be decreased.⁶ When multiple digits are significantly shorter than normal, deepening of the web spaces can achieve a relative increase in length and capacity for abduction. This can allow better stability in grasping larger spherical or irregularly shaped objects.⁶ When syndactyly is also present, the indications are even greater.

When multiple digits are more severely involved with a deficient bony structure, but have adequate soft tissue envelopes, toe phalanx bone grafting can be considered,^{5,69,71,86-90} particularly when there is adequate active flexion and extension. Even small flail nubbins may be useful to patients as they grow, such as to manipulate thread. Waiting before deciding the fate of such nubbins and adequate assessment by an occupational therapist are strongly recommended. If they interfere with function, they are best amputated. Amputation can often be combined with web deepening of the central portion of the hand,⁶⁹ achieving a relative lengthening of the remaining brachydactylic digits.

Rare cases of multiple digital brachydactyly can be appropriate for an "on-top-plasty" of one finger stump onto another (usually adjacent) stump.⁹¹ This technique is most useful to reconstruct the first web space components, such as transferring the index stump onto the middle finger stump, thereby creating a longer ulnar component and wider first web.^{6,91} Adequate bony elements and appropriate relative neurovascular pedicle lengths are essential in performing an on-top-plasty (that is, if the metacarpal of the digital stump being transferred is significantly shorter than that of the recipient, one may have to place the recipient joint or joints into flexion), to prevent a permanent flexion contracture.⁶ Distraction lengthening can facilitate some cases of on-top-plasty.⁹¹

If there is sufficient bone volume and length, various types of distraction lengthening^{5,92-99} may be helpful. It is nevertheless important to keep in mind that this option will not provide additional joints or increased motion and may even, in some cases, result in some stiffness of the

functional joints present.^{6,8,99} It is therefore best to reserve this technique for cases with functional loss of prehension and grip rather than for cosmetic reasons,⁹⁴ although some patients may desire surgery, after thoughtful discussion, for appearance only.⁹⁶ In cases of monodactyly or peromelia (that is, adactyly), free neurovascularized toe transfer^{32,71,88,100-102} may be an option to provide pinch and facilitate bimanual tasks.

Position and Marking

The patient is positioned supine, with the involved limb abducted and resting on a hand table. In very small patients, the head and shoulder can rest on the proximalmost part of a sufficiently wide hand table. The rest of the body is then positioned somewhat obliquely away from the hand table toward the opposite-side foot of the main operating table. This allows the hand to extend further on the hand table (see Fig. 47-8 in Chapter 47) to facilitate the surgeon's approach to the hand. When access to the foot or iliac crest is required, this position allows both to be reached. A tourniquet is applied and used intraoperatively on the upper limb, at 200 mm Hg or less in younger and 250 mm Hg or less in older healthy normotensive patients,^{103,104} and on the lower limb between 250 mm Hg or less and 300 mm Hg or less, respectively.¹⁰³ The tourniquet is deflated for 15 minutes or more¹⁰⁴ after 1.5 to 2 hours of inflation.^{103,104} According to Crenshaw,¹⁰³ it is less traumatizing to leave the tourniquet in place for 2 hours, or even a few minutes longer, and then deflate without reinflating it, than to deflate it for 10 minutes and reinflate it for the additional time required. Markings are made according to the technique planned (see the next section on Surgical Technique), and the patient is prepped and draped as usual. Prophylactic antibiotics are not generally used, unless there is a specific indication, such as prophylaxis for free toe phalanx transfer⁸⁶ or long operations such as free neurovascularized toe transfer.

SURGICAL TECHNIQUE

Web-Deepening Procedures

There are two general forms of web deepening: syndactyly releases, which are used between two short fingers that have a degree of syndactyly or between two short fingers that require relative lengthening; and multiple Z-plasties, which are used to deepen the space between a reduced number of fingers to allow a broader grip.

Syndactyly Releases

In cases of moderate brachydactyly or symbrachydactyly, standard syndactyly releases can sometimes be sufficient and beneficial (Fig. 48-9). The age at release is similar to that suggested for standard syndactylies. Some authors suggest treatment of the first web at or before 6 months of age to allow appropriate development of grasp between the thumb and the rest of the hand,¹⁰⁵ because patterns of hand function develop between 6 months and 2 years of age,^{105,106} and thumb opposition develops at approximately 7 months of age.¹⁰⁷ It is also suggested that surgery be undertaken before 1 year of age for any web where a difference in the length of the digits causes tethering.^{105,108} Otherwise, releases should be undertaken after 18 months of age to achieve the best results.¹⁰⁵ I often prefer to wait until a child is 9 to 12 months of age to release the first web, because anesthesia is less risky, surgery is easier, and neurovascular risk and the risk of flap necrosis are lower, as a result of the digital structures at this age being somewhat larger. Unless a release is required at an earlier age, for any of the previously mentioned reasons, I also prefer to perform surgery for the rest of the webs after 3 years of age, when collaboration with postoperative therapy—that is, the ease of pulling on compressive gloves, and keeping on silicone

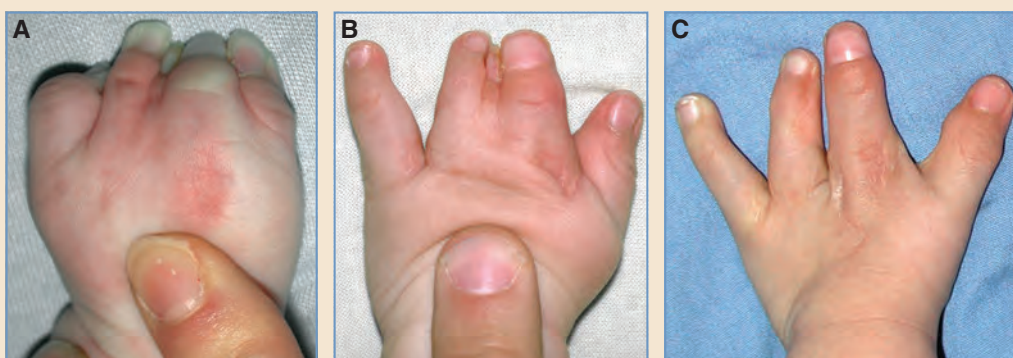


Fig. 48-9 Syndactyly releases in the treatment of symbrachydactyly. **A**, Left hand before surgery. **B**, Left hand after combined first and fourth web syndactyly release, which were performed together, because eventual amputation of the third severely hypoplastic ray was planned, permitting the correction of the web between the second and fourth digits to be performed at that time. **C**, Left hand after amputation of the third ray and completion of all syndactyly releases.

and splints—is much easier. All necessary releases should nevertheless be completed by school age and planned accordingly. If all four web spaces require release, the first and third should be done simultaneously¹⁰⁵ at an early enough age to allow appropriate cortical integration of the thumb. The second and fourth webs can then be operated on together,¹⁰⁵ after the scars from the first operation have sufficiently matured and softened to allow reintervention. If only the second through fourth web spaces require release, the second and fourth should be done together first, followed by the third.¹⁰ Most authors suggest not operating on both sides of the same digit at the same sitting,¹⁰⁵ to avoid jeopardizing the vascular supply to the digit.¹⁰ A technique involving three adjacent web spaces has nevertheless been described.¹⁰

When several adjacent web spaces require correction, it is important to plan as if each digit will end up with only one arterial pedicle. This approach is important, because the division of the common digital artery into proper digital arteries may be more distal than usual^{4,69} and because relative lengthening of short digits often requires ligation of one of the two digital arteries, even if their bifurcation is at the usual level. During surgery, the two nonadjacent web spaces are explored, once the syndactyly release incisions have been made, to ensure that none of the proper digital arteries is absent. The proper digital artery that one wishes to sacrifice in each space is clamped, and the tourniquet deflated to verify the blood supply to each digit. When this is done, one must ensure that, should one of the two arteries require ligation in the intervening web space, the vascular supply of the corresponding digit is intact on its opposite side.

Because these digits are shorter than normal, I use two relatively short, proximally based triangular flaps, one based on the palmar side and the other on the dorsal side of the syndactyly, to line the depth of the web³² (Fig. 48-10). This decreases the quantity of the skin graft necessary on the adjacent digital bases compared with a single proximally based rectangular flap used to line the depth of the web. There are authors who still prefer the use of a wide dorsal flap for their syndactyly releases to prevent the development of a narrow V-shaped commissure.¹⁰⁵ When used, the two triangular flaps are drawn slightly offset in a radioulnar direction, one in relation to the other, and long enough so that their tips cross over somewhat. Full-thickness skin grafts are used to close the remaining open wounds to limit contraction. Split-thickness grafts are avoided, because they often result in recurrence of the syndactyly. The donor site for a smaller graft can be the proximal ulnar forearm or the palmar non-hair-bearing distal forearm (harvested in a lon-



Fig. 48-10 Syndactyly release for symbrachydactyly. **A**, Left hand, dorsal aspect, before surgery. **B**, Left hand, palmar aspect, before surgery. **C**, Left hand, dorsal aspect, at completion of surgery. Note the use of relatively short triangular proximally based dorsal flaps to help line the depth of each web, the tips of which overlap side to side with the corresponding palmar short triangular proximally based flaps. **D**, Left hand, palmar aspect, at completion of surgery.

gitudinal direction to avoid the social stigma associated with transverse wrist scars), with a very good color match in most patients. For larger grafts, the infrequently hair-bearing lateral lower abdominal skin can be used. Despite larger quantities of skin, this lateral lower abdominal skin is not as good of a color match for the hand. The inguinal skin is not only a poor color match, becoming darker in more pigmented skin types,¹⁰ but also often grows hair after puberty. The Sherif technique,¹⁰⁹ which usually does not require skin grafts when used for simple syndactyly release, is indicated only when a single web requires treatment. Transverse skin laxity on the dorsum of the hand rarely allows the harvesting of several adjacent Sherif flaps, in addition to leaving conspicuous longitudinal scars.

Multiple Z-plasties

In cases where the digital nubbins (which replace the index, middle, and ring fingers), require amputation, multiple serial Z-plasties^{69,110} should be considered to deepen the space between the hypoplastic thumb and the little finger (Fig. 48-11). This should be done before the child is of school age, and when collaboration with postoperative therapy is sufficient.

At surgery the incisions are drawn, but the flaps are not all incised at once. Starting at one end of the planned web, the first pair of flaps is incised, interposed, and tacked before moving on to the next set of flaps, and so on. This avoids errors in flap design, because when one moves toward the depth of the web, the flap angles change from what was originally drawn. It may be

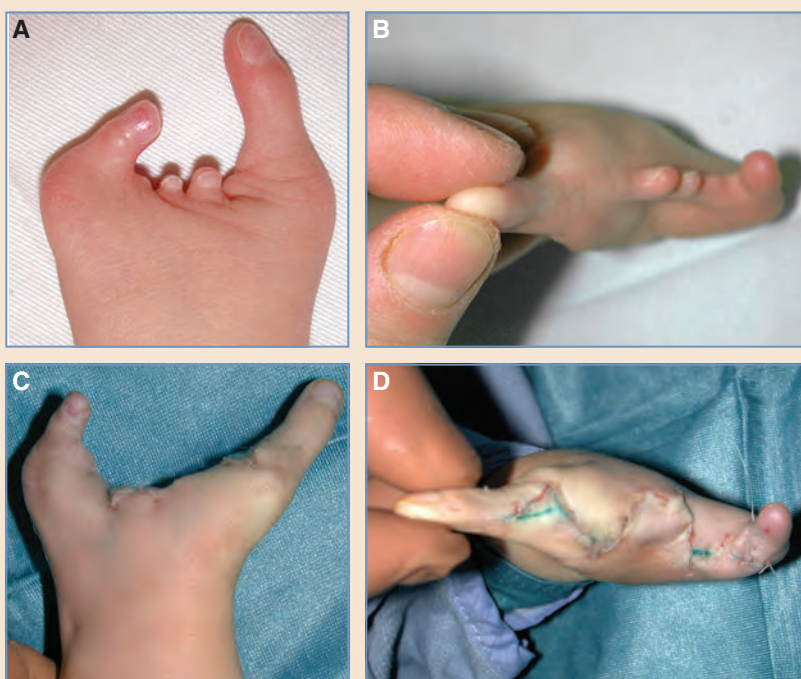


Fig. 48-11 Multiple Z-plasties to deepen the web between the only two useful border digits in symbrachydactyly of a patient's left hand. **A**, Dorsal preoperative appearance. **B**, Web preoperative appearance. **C**, Dorsal appearance at the end of surgery; amputation of useless vestigial nubbins was undertaken simultaneously. **D**, Web at the end of surgery; interdigitating flaps decrease the risks of web contracture formation.

useful to shorten and contour the distal ends of the hypoplastic metacarpals present in the depth of the new interdigital space and/or excise any extremely hypoplastic rays that cannot benefit from reconstruction.¹¹⁰ In some cases shortening the metacarpals may be contraindicated, because they provide increased grasp stability, and should therefore not be performed. Thorough preoperative occupational therapy evaluation can identify which patients should and which should not have their metacarpals shortened.

Bone-Lengthening Techniques

Lengthening techniques include free toe phalanx transfer, on-top-plasty, ray transposition, distraction, and free neurovascularized toe transfer.

Free Toe Phalanx Transfer

If the cutaneous sleeve is of sufficient dimensions, an appropriate reconstructive method (Fig. 48-12) includes the transfer of a free, nonvascularized toe phalanx. The toe phalanx allows a better long-term survival rate, approaching 100%, unlike conventional bone grafts from the iliac crest or fibula, which resorb.³² To have a chance of surviving, the bone graft must include the periosteum.*

*References 5, 6, 10, 32, 69, 71, 86, 87, 89, 90.

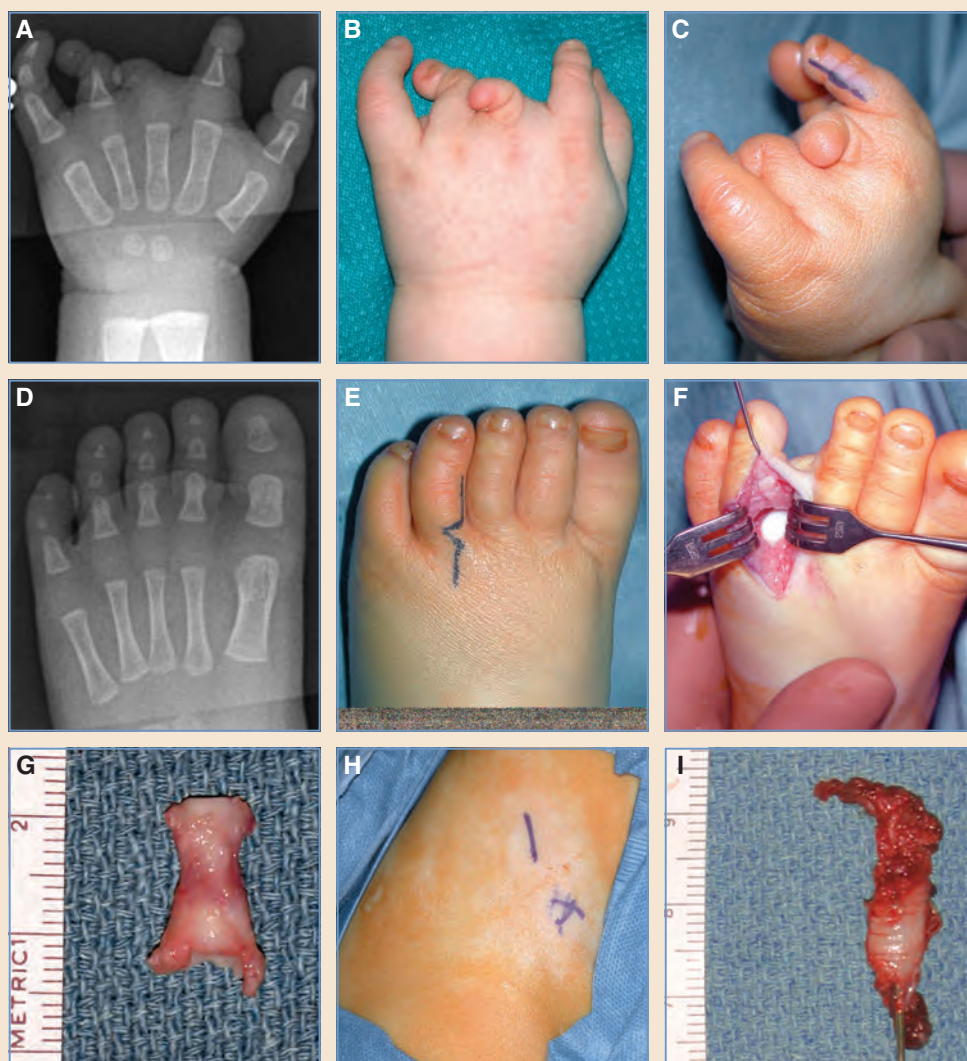


Fig. 48-12 Free toe phalanx transfer to the index finger in symbrachydactyly. **A**, Preoperative posteroanterior radiograph of the left hand. **B**, Preoperative appearance of the left hand. The index fingertip is unstable for pinch. **C**, Planned incision for the free toe phalanx transfer to the monophalangeal index finger to stabilize the flail tip. **D**, Anteroposterior radiograph of the left foot in which the fourth toe proximal phalanx will be harvested. **E**, Planned incision for harvesting the fourth toe proximal phalanx. **F**, The fourth toe proximal phalanx has been removed; the extensor tendon is retracted to the lateral side and the head of the metacarpal is visible in the depth of the wound. **G**, The fourth toe proximal phalanx has been harvested including the periosteum, proximal collateral ligaments, the plantar plate, and the proximal joint capsule. Unfortunately, despite precise measurements on the preoperative radiographs, the width and length of the phalanx were excessive when attempting its transfer to the index finger and required reduction. During the reduction procedure, the periosteum was kept pedicled to cover the sites of bony reduction. **H**, The right iliac crest was prepared for harvesting a bone graft to fill the donor defect in the toe. (**NOTE**: X marks the anterosuperior iliac spine. The incision is planned for just below the iliac crest to prevent a scar in the area subjected to the most friction from clothing, and posterior to the X to avoid branches of the lateral femoral cutaneous nerve.) **I**, Bone of adequate width and length for the donor defect in the toe, is harvested from the iliac crest with a trepan.

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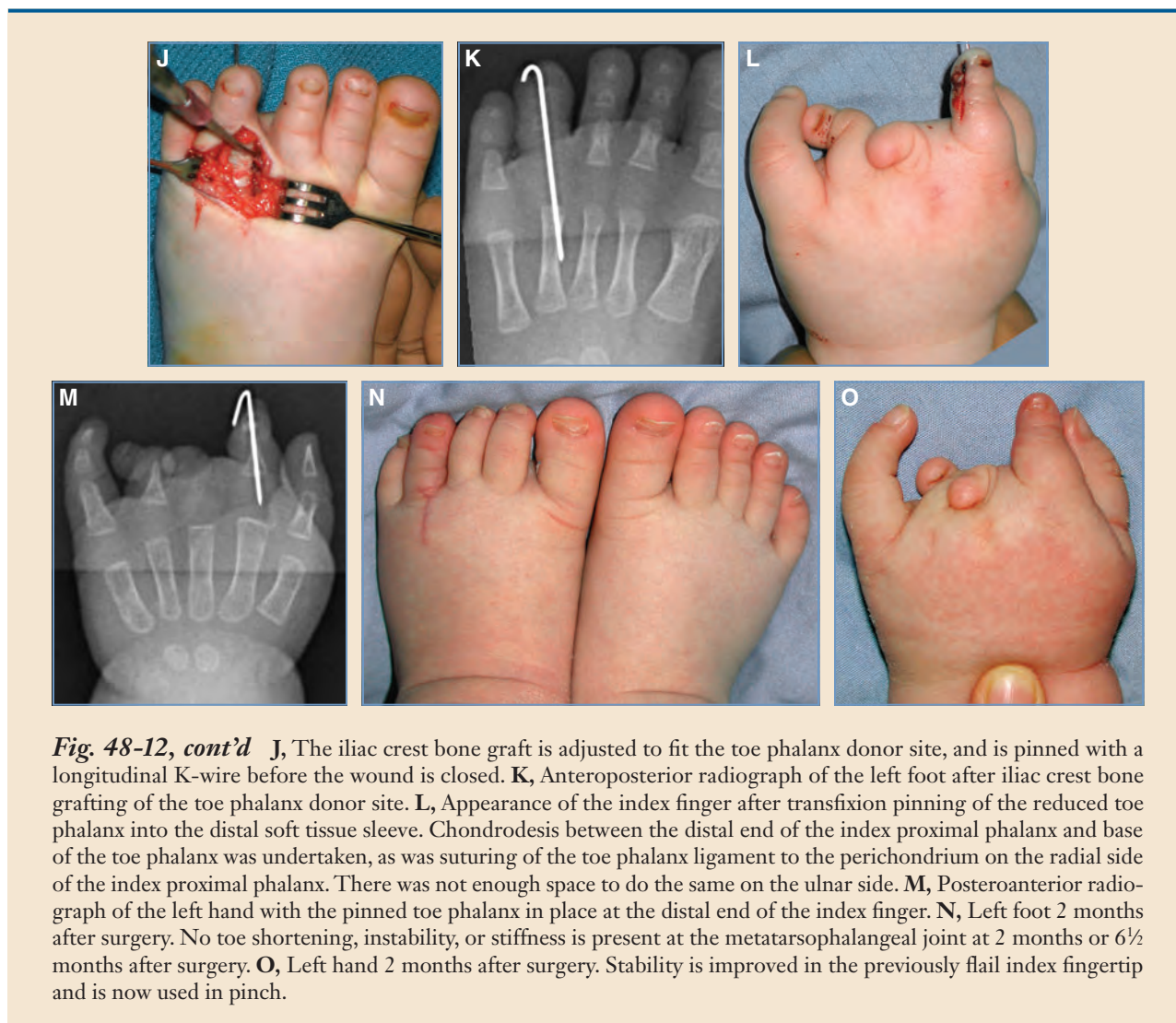


Fig. 48-12, cont'd **J**, The iliac crest bone graft is adjusted to fit the toe phalanx donor site, and is pinned with a longitudinal K-wire before the wound is closed. **K**, Anteroposterior radiograph of the left foot after iliac crest bone grafting of the toe phalanx donor site. **L**, Appearance of the index finger after transfixion pinning of the reduced toe phalanx into the distal soft tissue sleeve. Chondrodesis between the distal end of the index proximal phalanx and base of the toe phalanx was undertaken, as was suturing of the toe phalanx ligament to the perichondrium on the radial side of the index proximal phalanx. There was not enough space to do the same on the ulnar side. **M**, Posteroanterior radiograph of the left hand with the pinned toe phalanx in place at the distal end of the index finger. **N**, Left foot 2 months after surgery. No toe shortening, instability, or stiffness is present at the metatarsophalangeal joint at 2 months or 6½ months after surgery. **O**, Left hand 2 months after surgery. Stability is improved in the previously flail index fingertip and is now used in pinch.

Most authors recommend using the fourth or possibly the third toe,⁹³ and keeping the second toes and possibly even the third toes, for possible future free neurovascularized transfer. The fifth toe is avoided because of potential problems with donor site instability,⁸⁶ particularly when the donor site is not bone grafted. The proximal⁸⁹ or middle⁸⁶ phalanx can be used. An anteroposterior radiograph of the toes permits rough measurements to be obtained,⁸⁶ helping to determine which phalanx would be the most appropriate.

This technique is most appropriate when there is good active flexion and extension of the nubbin, and when pinch or global hand function will improve significantly as a result of this additional stable digit.⁷¹ When grafting a monophalangeal digit in which functional tendons are present and can mobilize the proximal phalanx, the transfer is even more functional (J. Upton III, personal communication, 2006).

The recommended age at surgery varies somewhat. Buck-Gramcko⁸⁷ noted that after surgery between 7 and 18 months of age, the growth plates remain open in the highest percentage of grafts (65.7%) compared with older patients, whereas between 1.5 and 4 years the average growth is the greatest (3.5 mm) compared with either the younger or older age groups. In the

series undertaken by Cavallo et al,⁸⁶ average growth was also somewhat better in the middle age group (that is, 18 months to 4 years), although it was more modest, with an average of 0.2 mm of growth, whereas the younger and older age groups each had a net average resorption of 0.4 and 0.1 mm, respectively, at follow-up. Radocha et al⁸⁹ obtained physeal openness in toe phalangeal grafts of 94% in those operated on before 1 year of age and 71% for those operated on between 1 and 2 years of age, with a severe drop-off thereafter. They measured growth to be, on average, 1.0 mm per year for both of these groups and less so in the older children. Unglaub et al⁹⁰ noted the least resorption (9%) and the most growth (73%) in the youngest age group operated on between the ages of 6 and 18 months compared with the older patients. These findings did not correlate with the presence of open growth plates at follow-up examination (on average, 3.5 years after surgery), which were 73% in the youngest age group, 90% in the children between the ages of 18 months and 4 years, and 71% in the older patients.⁹⁰ Dobyns⁵ suggests that surgery be performed before 18 months of age, Kay and McCombe⁷¹ recommend surgery only for patients 15 months of age or younger, whereas Peimer and Alexander⁸⁸ suggest that this operation be performed before 1 year of age. Of note, true growth is a difficult parameter to evaluate in a growing child. Its measurement is subject to many variables, including variations in radiographic magnification (dependent on the distance between the bone and the radiographic plate), the increasing degree of bone calcification over time, and the growth of the bone relative to the bones of the normal hand, the donor foot, and the other bones of the affected hand.

In the hand, a longitudinal palmar,⁷¹ dorsal,^{71,86,89} lateral (J. Upton III, personal communication, 2006), dorsal zigzag,^{6,87} dorsal curved,⁶ or dorsal V-Y incision⁸⁸ is made. In multiple proximal digit grafting, a transverse dorsal distal metacarpal incision (J. Upton III, personal communication, 2006) can be used instead. The soft tissue envelope is gently handled throughout the operation, and some fat and fibrous tissue is removed to accommodate the planned phalanx transfer.^{71,87,88} The skin at the tip of the soft tissue sleeve must remain intact and well vascularized.⁶ The size of the available space can be assessed by inspection, and this, along with measurements on the preoperative radiographs of the toes, can help determine which phalanx is the most appropriate for the transfer.⁸⁶ It is important to remember that significant portions of the toe phalanx are cartilaginous at this age and therefore not visible on the radiographs. Cavallo et al⁸⁶ also noted that complete harvest of the second phalanx seems more resistant (less resorption [3 mm versus 6 mm]) and shows more growth (2 mm versus 1 mm) than subtotal harvest of the first phalanx on follow-up examination after transfer. It is therefore probable that completely harvesting a phalanx of the appropriate size, rather than shortening one that is too long, will give better results.

The selected toe phalanx is approached either through a dorsal longitudinal,^{5,86,89} zigzag,^{71,87} S-shape,⁶ curvilinear,⁶ or midlateral incision (J. Upton III, personal communication, 2006). The extensor tendon is retracted to one side,^{6,87} or split^{5,6,70,71,89} and elevated from the underlying periosteum. The flexor sheath is released to free the flexor tendon from the overlying bone (J. Upton III, personal communication, 2006).⁷¹ To ensure the best growth plate survival and subsequent growth, the chosen phalanx is harvested including all its periosteum,* capsular attachments,^{5,6,71} proximal collateral ligaments,^{5,6,32,87,89} and in the case of some investigators, plantar plate.† Occasionally, if joint reconstruction is indicated, the head of the proximal phalanx, along with the proximal interphalangeal joint and the entire second phalanx, can be harvested.⁷¹ The donor defect can be treated in several different ways. Some leave it to retract, after repair of the extensor tendon, if split, and after closure of the skin.^{6,86} Others stabilize the defect with a longitudinal K-wire.^{71,89} Some investigators fill the donor defect by suturing the extensor, flexor, and what is left of the collateral ligaments together to serve as a spacer.^{87,111} Finally, others graft the site with the use of a cylindrical piece of iliac crest bone, capped with epiphyseal cartilage, which

*References 5, 6, 10, 32, 69, 71, 86, 87, 89.

†References 5, 6, 32, 71, 87, 89.

is placed proximally and immobilized with a K-wire (J. Upton III, personal communication, 2006).⁷¹

In the hand there is a coalescence of fibrous tissue, including attachment of the flexor and extensor tendons into a “cap.” Some leave the cap intact, suturing the phalanx transfer to its distal surface.⁸⁶ Others prefer to either divide this cap transversely into palmar and dorsal components to reconstruct the flexor and extensor tendons,⁸⁷ sagittally to reconstruct the collateral ligaments,⁸⁹ or in a cruciate fashion into palmar, dorsal, radial, and ulnar components.⁷¹ When reconstructing the collateral ligaments, the radial and ulnar components are used, whereas the palmar and dorsal components are used for the tendons. Because of the relatively greater strength and excursion of the flexor tendon, it is best to leave the flexor tendon relatively longer than the extensor tendon when dividing the cap to prevent a flexion contracture. If there is a sufficient amount of bone in the digital proximal phalanx, the tendons present are already inserted into the base of this phalanx.⁷¹ In this case, the distal end of the digital proximal phalanx and the base of the toe phalanx transfer only require preparation for either chondrodesis⁷¹ or bone-to-bone fusion while preserving the epiphysis.⁸⁷

The toe phalangeal transfer is inserted into the recipient pocket in the hand, ensuring that tension on the skin does not cause ischemia.⁶ The transfer is immobilized with a K-wire,^{5,71,86-89} while minimizing trauma and heat to avoid injury to the epiphysis.⁷¹ The ligaments can be repaired,^{71,87-89} while the extensor tendon is reattached to the base of the toe phalanx^{87,89} or to the dorsal capsule of the toe phalanx transfer,⁷¹ when indicated. The flexor tendon, where appropriate, is either attached to the palmar base of the toe phalanx transfer, leaving the palmar plate free to attach through adhesions⁸⁷ or to the palmar plate itself.^{71,89} Skin closure may be done first, but if there is tension, a Z-plasty,⁷¹ local rotation flap,^{87,89} or transposition flap⁸⁷ may also be necessary.

On-Top-Plasty

On-top-plasty^{6,91} is rarely used because of the risks of circulatory compromise in the transferred segment or a flexion contracture of the transferred segment as a result of the positioning necessary to reduce the tension on its neurovascular pedicle. It should be considered only in selected cases when these risks are low. On-top-plasty is most often indicated to reconstruct the first web space components such as transferring the index stump onto the middle finger stump, thereby creating a longer ulnar component and wider first web,⁶ or onto the first ray to reconstruct a hypoplastic thumb.^{5,91} An example of the former is the presence of a second metacarpal, with or without a small, actively mobile portion of the proximal phalanx, too short and too close to the thumb for use in functional pinch, adjacent to a relatively shorter third metacarpal. The second ray is dissected as a neurovascularized pedicled finger ray, similar to the manner undertaken in pollicization, and transferred onto the third metacarpal base. The base of the second metacarpal may be left behind, if its inclusion would result in excessive neurovascular tension. The first web is thereby deepened, the third ray is lengthened, and the longer digit is in a better position for pinch with the thumb. Although there is no recommended age for this procedure in the literature, there may be some benefit in waiting until the patient is less prone to vasospasm, which is after 1 year of age or more than 10 kg in weight (J. Dubois, personal communication, 2006). Occasionally it may be helpful to distract an otherwise useless segment with the aim of transposing it as an on-top-plasty to a more useful position.⁹¹

Ray Transposition

Another ray transposition, although not a true on-top-plasty, has been described by Kay and McCombe.⁷¹ In a case of monodactyly, the thumb, with no mobility at any joint, was transposed to the ulnar side of the hand as a neurovascular pedicled transfer. This was done in preparation for a free neurovascularized transfer of the mobile second toe to the thumb position to improve grasp. A similar technique, but employing a microsurgical free transfer, can be used for transfer-

ring an otherwise useless radial finger vestige into an ulnar position or as an on-top-plasty onto an ulnar metacarpal remnant.^{5,32}

Distraction

There are three variations on distraction: distraction with immediate bone grafting, gradual distraction with secondary bone grafting, and gradual distraction with callotasis bone formation. Distraction with immediate bone grafting permits more conservative lengthening in a single stage. Gradual distraction with secondary bone grafting allows greater lengthening but requires bone grafting at completion. Gradual distraction and callotasis bone formation has the advantage of achieving greater lengthening without necessitating bone grafting in successful cases, but it requires additional months of immobilization with a risk of decreased resistance to stress.⁹⁹

Distraction With Immediate Bone Grafting

Distraction with immediate bone grafting results in limited lengthening of certain long bones, but only if there is enough bone stock to allow for osteotomy, handling, fixation, and bony consolidation. The age at surgery can vary from preschool age, recommended for its positive psychological benefit, to when a patient is old enough to decide for himself or herself.⁹ Although there is no clear recommended age for this procedure, the older the patient, the more likely it is that the previously mentioned preoperative requirements are met. In a report by Saito et al,⁹⁸ two patients were 13 years of age at the time the procedure was performed and the age of the third patient was not mentioned.

Joint contractures are a risk with any type of distraction lengthening.^{6,8} One-stage lengthening with bone grafting is rarely advocated in the phalanges, for fear of upsetting the delicate balance between the flexors and extensors; therefore one-stage lengthening is mostly used to lengthen metacarpals.⁵ Although most authors^{8,39} use a dorsal approach for metacarpal lengthening, Saito et al⁹⁸ described a palmar approach. In addition to a transverse osteotomy,¹¹² various other bone cuts have been described to improve stability, including a step cut,¹¹³ a V shape,^{39,98} a chevron shape,¹¹⁴ and a dowel shape, which is a piece of iliac crest in which holes are reamed to support the two ends of the lengthened metacarpal.¹¹⁵ Soft tissue release, muscle slide, and reconstruction of the intermetacarpal ligament are often necessary to achieve maximum lengthening.^{6,39,98,112,114} Reattachment of the interosseous muscles to the bone is advocated by some after muscle slide.^{6,39} An iliac crest bone graft is often used,^{6,39,98,114,115} although fibula and other sources of bone grafts have been advocated.⁶ Elongation with the use of this technique is between 5 and 15 mm,^{9,98,116} and fixation is achieved by means of intraosseous wires,⁹ K-wires,^{5,9,39} plates and screws,^{5,39} a K-wire-fixed distraction device,¹¹³ or an external fixator.⁸

Gradual Distraction With Secondary Bone Grafting or Callotasis Bone Formation

Distraction lengthening generally necessitates sufficient bone to permit stable insertion of the fixation device in addition to sufficient bone necessary for the osteotomy, handling, and bony consolidation. These details therefore affect the age at which distraction is undertaken. Some authors state that distraction should be postponed until 1 or 2 years of age.¹¹⁶ Others believe that because of the size of most distraction devices, distraction is impractical before 2 years of age, but should be completed by the age at which school begins⁶ or even between 2 and 4 years of age.³⁹ Others recommend, for brachymetacarpia at least, waiting until the patient is 8 to 14 years of age¹¹⁶ or in the range of 10 to 15 years of age because of excellent callus formation, consolidation, and near closure of the growth plates, simplifying estimation of necessary lengthening; in addition, patient motivation is higher in this older age group and patients can adequately care for the fixator by themselves.⁹⁵

Despite the varying opinions, the age at which surgery has been performed varies widely in the literature. Arslan's patients ranged from 10 to 14 years of age,⁹² Kato et al's⁹⁵ from 10 to

19 years, Belusa's patient was 16 years of age,⁹³ the patients of Pensler et al⁹⁷ were from 2.5 to 7 years, those of Dhalla et al⁹⁴ from 2 to 16 years, and of Matsuno et al⁹⁶ from 1 to 13 years of age. Dhalla et al⁹⁴ and Matsuno et al⁹⁶ used variations in technique that permitted very small bones to be addressed, making it possible to treat some very young patients. Nevertheless, this technique is usually recommended for larger bones. Both Dobyns⁵ and Flatt⁶ suggested limiting its use to bones of at least 3 cm in length. Double pin fixation on either side of the osteotomy is generally suggested to maintain alignment during distraction.⁸ Using this technique, Dhalla et al⁹⁴ recommended a minimum bone length of 2.3 cm but as little as 1 cm using a modification with a single pin on either side of the osteotomy along with a longitudinal intramedullary K-wire to provide stability. Even smaller bones, as short as 3 mm, have been successfully lengthened by Matsuno et al⁹⁶ using an apparatus that could fix transverse K-wires—placed across joints where necessary—along with a longitudinal intramedullary K-wire to increase stability. Some authors use a longitudinal intramedullary wire, during metacarpal lengthening, through the metacarpal and phalanges to maintain alignment, even with larger bones.^{39,116} Others use longitudinal K-wires for a different reason, pinning joints distal to the area of distraction to avoid progressive flexion contractures common to lengthening.^{6,88,116} External splinting has also been used for this reason.⁶

Although lengthening by gradual distraction has been used for both phalanges and metacarpals, only 23 of the 75 bones elongated by a number of different investigators were phalanges.⁹²⁻⁹⁷ It is important to note that this technique does not create new joints, thus excessive elongation can result in a stiff,^{8,99} thin,⁹⁹ or even contracted⁸ digit. Some authors therefore believe that the best candidates for phalangeal distraction are those who have significant shortening along with an associated deformity, such as angulation, that requires correction,⁸ or occasionally, to create some form of pinch where it is absent. Both single and multiple digits can be treated,⁶ even simultaneously in the same hand.^{88,95-97} As previously noted, it may be useful to consider distracting an otherwise useless segment with the aim of transposing it as an on-top-plasty,⁹¹ usually on either side of the first web.^{5,91}

The distraction technique involves access through a longitudinal incision usually on the dorsum of the hand (Fig. 48-13) for the metacarpals,^{8,39,94,95,116} which is slightly offset in relation to the axis of the digit.^{95,116} An osteotomy is made, midway between sets of double (or single⁹⁴) half-pins^{8,95} or K-wires inserted to stabilize the proximal and distal fragments.^{5,6,39,116} Some prefer to make the osteotomy in the metaphysis,³⁹ whereas others prefer the middiaphyseal region⁹⁴ or even through a joint system.⁵ The cut is made either through both periosteum and full-thickness bone, which usually requires bone grafting at the end of distraction^{5,6,39,99,116} or preserving the periosteum.^{39,94,95,97} A step-cut osteotomy in the diaphysis can occasionally allow distraction without necessitating either callotaxis bone formation or bone grafting, permitting consolidation between the surfaces of the bone in contact at the end of distraction (see Fig. 48-13).

As previously mentioned, a longitudinal intramedullary K-wire can be inserted in the metacarpal and phalanges to maintain alignment, as well as fixation of the proximal and distal fragments.¹¹⁶ Various types of frames exist, including K-wires mounted in a half-circular frame,¹¹⁷ K-wires mounted in a bilateral longitudinal frame,^{5,39,116,118} and half-pins mounted in a unilateral longitudinal frame.^{94,97} The Ilizarov method, which also includes the preservation of the medullary cavity, is not practical in the small bones of the hand.³⁹ Multilayer closure is undertaken, a dressing applied, and distraction begun in the days after surgery, according to the surgeon's protocol. In cases where there are problems with consolidation^{91,95} or when consolidation is not expected at the end of distraction, the gap is grafted with an iliac bone graft,^{6,39,95,99} toe phalanx,^{6,39} fibula, bone from a vestigial digit, or other source.^{6,39,99,116} Most patients (71 out of a combined 75 patients) go on to form bone through callotaxis, healing without the need for secondary bone grafting.⁹²⁻⁹⁷

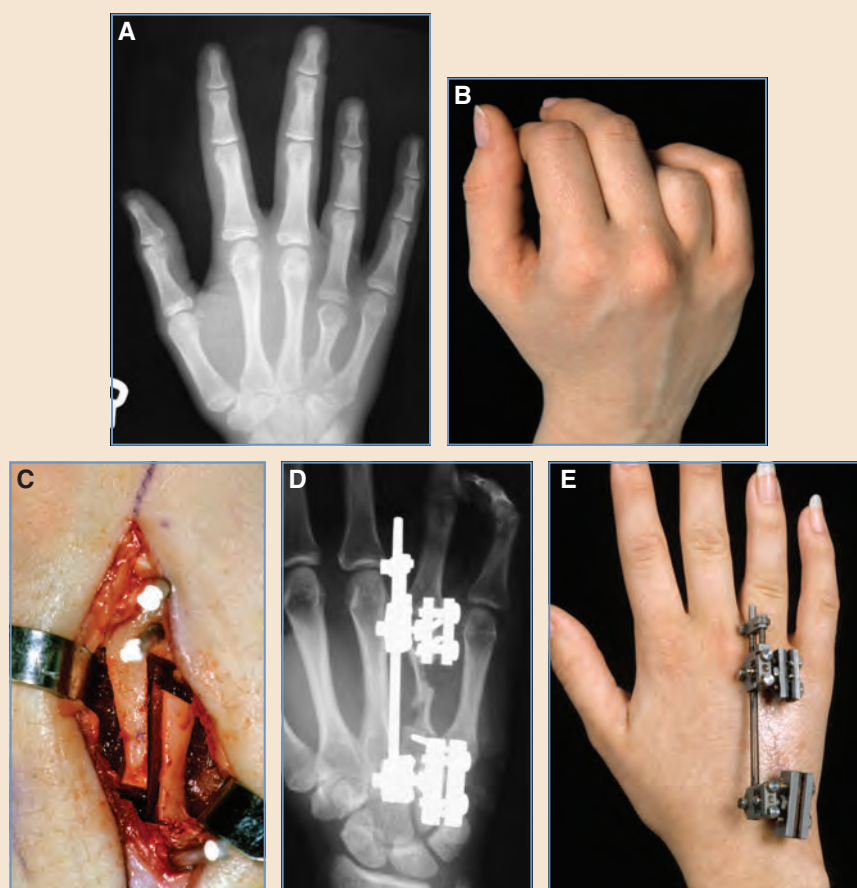


Fig. 48-13 Distraction lengthening for brachymetacarpia of the right fourth metacarpal. **A**, Preoperative posteroanterior radiograph of the right hand. **B**, Preoperative view of the right hand. **C**, In this case, a step-cut osteotomy was made, through a dorsal longitudinal incision, to permit consolidation without bone grafting or callotasis bone formation. Two sets of half-pins were used on either side of the osteotomy for fixation. **D**, Right hand posteroanterior radiograph during distraction lengthening. Some deformity into flexion occurred during distraction, necessitating realignment in the operating room. **E**, Right hand during distraction lengthening.

Free Neurovascularized Toe Transfer

Finally, there is the possibility for free neurovascularized toe transfers to the hand (Fig. 48-14). Some authors suggest that this procedure be performed at 2 years of age.¹¹⁹ Others suggest that surgery be performed at an even younger age, because patterns of hand function are generally established between the ages of 6 months and 2 years,¹⁰⁶ with thumb opposition beginning at approximately 7 months of age.¹⁰⁷ Lister¹⁰² recommends that in unilateral deformities, the surgery be completed before 18 months of age, but mentions that it can be undertaken as early as 6 months of age. Nevertheless, some prefer to wait until after the patient reaches 4 or 5 years of age⁸⁸ before undertaking such transfers. In bilateral deformities, the transfer will probably be used regardless of the age at which surgery is undertaken.^{101,102}



Fig. 48-14 Free neurovascular transfer of a second toe to the ulnar side of the hand for monodactylic aphalangic symbrachydactyly. **A**, Preoperative anteroposterior radiograph of the palmar surface of the right hand. **B**, The palmar surface of the right hand before surgery. **C**, The palmar surface of the right hand during surgery, after web deepening and preparation of the recipient site on the ulnar side, including the identification of appropriate vessels, nerves, and tendons. **D**, Preparation of the left second toe for free neurovascularized transfer. **E**, Appearance of the foot donor site at the end of surgery after closure of the defect. **F**, Appearance of the well-vascularized transferred toe in the dressing at the end of surgery. **G**, Use of the transfer 3 years postoperatively.

The second toe (or toes) in children are usually used to minimize the donor defect in the foot.^{71,101,102,120} Rarely, a second and third toe^{32,121} en bloc transfer or even a combined third and fourth toe transplantation¹²¹ have been undertaken. Others consider these combined toe transfer procedures too disfiguring¹⁰⁰ and have found them to cause, at least in adults, more foot problems than a single first or second toe transfer.¹²⁰ Rarely, other types of transfers, such as a wraparound free flap or a partial toe transfer, may be indicated in select cases in the older child.⁷¹ Toe transfers are generally not a solution for replacing one, two, or even three missing fingers, because they do not resemble or function like normal digits and their range of motion is frequently limited.^{32,71,101} There are, nevertheless, cases in which reconstruction using toe transfers improves prehension patterns and appearance by giving the hand a total of two or three “fingers.”^{32,100,119,121} This is achieved by transferring a single second toe or two second toes, or performing a double toe transfer, to a hand that has a single finger or no digits at all.

Although toe transfers are most often used to replace a missing thumb,^{71,100,101} in *syndactyly*, the thumb is usually the last digit absent. If this thumb is short, unstable, or poorly oriented, the previously mentioned lengthening and other standard stabilization and reconstructive procedures are indicated. These more classical methods may also be used to provide an adequate ulnar post against which a stable, well-positioned thumb is able to grasp. Otherwise, toe transfers can replace an absent ulnar post and/or thumb. The best results are obtained when the metacarpophalangeal joint of the finger (or fingers) is present⁷¹ and, in the thumb, when the carpometacarpal joint is stable and mobile.^{71,100} If the metacarpophalangeal joints are absent or unusable, a chondrodesis or arthrodesis between the proximal phalanx of the transfer and the hand metacarpal is generally more stable and allows better orientation of the digit.⁷¹ If the thumb basal joint is present, and two toe transfers are planned, some authors suggest that one toe be positioned in the thumb location and the other on the third or fourth ray.⁷¹

The more proximal the digital absence, the more likely it is that necessary recipient structures are inadequate.⁷¹ It is therefore essential to explore the recipient structures before harvesting the toe (or toes) and to determine the amount of bony framework (that is, whether or not the metatarsophalangeal joint should be included), length of tendons, neurovascular pedicles, and skin necessary for the intended reconstruction.¹²⁰ If the thumb basal joint is inadequate, some authors suggest that two relatively short toe transfers be placed, facing each other, close enough to permit the tips to touch in maximum flexion.^{71,100} This allows grasping of small objects, which is preferred to the ability to hold large objects.^{71,100} Others contend that, in unilateral cases, fine manipulation will be preferentially undertaken with the normal hand, so a strong grasp with a wide span should be reconstructed in the affected hand.¹⁰¹ Some have even undertaken the reconstruction of the basal joint by including the metatarsophalangeal joint in the free neurovascularized transfer.¹²² When the basal joint of the thumb is normal, the position is adjusted to permit both tip-to-tip and tip-to-side pinch.¹⁰⁰

Büchler³² has suggested some rather aggressive procedures in cases of *peromelic syndactyly* (that is, the absence of all digits at the metacarpal level). These include transferring one second toe to the thumb position and a double second and third toe to the third and fourth ray positions. He has also reported transferring the index finger from the normal hand to the third metacarpal and a double toe transfer to the fourth and fifth rays in a single sitting, followed 6 months later by a single toe transfer to the thumb position.³² Others prefer not to transfer more than two toes, and thus suggest two-toe hands in *peromelic* cases, or three-digit hands in *monodactylic* hands (including two toes and the thumb present), augmenting the thumb ray, if necessary, with a free phalangeal transfer.¹⁰⁰ If there is a clinical indication for lower limb amputation, transplantation of any useful toes should be considered.¹²³ Details of free neurovascularized toe transfer can be found in Chapter 51.

Joint Stabilization Operations

Rarely, unstable joints may be appropriate for formal ligament reconstructions³² (see Chapter 44). Many ligament reconstructions are best treated with joint fusion,³² particularly in cases where hypoplastic digits have too much laxity and underdevelopment of the supporting structures to achieve sufficient stability with other methods.

Other Corrective Procedures

Other corrective procedures include various techniques for improving function and/or appearance. Some examples are pronation osteotomy of the first metacarpal³² to improve the position of the digit for grasp, and tendon transfers to increase adduction or palmar abduction of the thumb.³² Amputation is appropriate in certain cases, such as when the following is present: vestigial nubbins⁶ that provide no increased function, are in the way, and are too small to accommodate toe phalanx transfers (see Fig. 48-11); portions of nonfunctional digits (see Fig. 48-9), parts of which can be used elsewhere⁸⁸; or a flail single hypoplastic ray in a normal or near-normal hand⁸ that interferes with function.

POSTOPERATIVE CARE

At the time of surgery, it is possible to increase postoperative comfort by using a digital or distal regional bloc, such as a median nerve or ulnar nerve at the wrist, with a long-acting agent such as bupivacaine. A well-padded dressing avoids pressure points. The dressing must not be too tight, which could cause postoperative compression and go unnoticed when a regional block is used, although this is less likely to occur than with a more proximal regional block. When the dressing is being applied, it is important to take into account postoperative swelling, which may be substantial if the total tourniquet time is more than 2 hours or if the surgery is extensive. Postoperatively, most upper limb procedures benefit from elevation to minimize edema. To achieve this, suspension can be used (see Chapter 47), and it is usually continued for approximately 1 to 2 weeks. It may be prolonged for procedures that are more extensive or if edema is pronounced. Subsequently the end of the sleeve is tied over the opposite shoulder to the back portion of the attachment around the chest, elevating the hand to the shoulder level. This arrangement is kept under the patient's shirt to help keep the dressing clean and intact.

After the appropriate duration of immobilization, occupational therapy begins. As already specified in Chapter 47 this includes, as required, splinting, scar massages, use of topical silicone to help control hypertrophic scars, compressive gloves (as used in burn scar treatment), exercises, and so on. Not all patients tolerate topical silicone, and some develop an eczema-like rash. In my experience, most patients will tolerate either shorter periods of topical silicone use, such as a few hours per day, or changing to one of the other products that are available. Before the use of topical silicone is abandoned completely, several different types should be tried, because the advantages far outweigh the associated problems. The patients are followed until the scars have matured, which may take 1 to 2 years and sometimes longer. Subsequently the patients are followed for variable amounts of time in the outpatient clinic, depending on the initial malformation, type of reconstruction, results, and the patient's and/or family's understanding of concerns that require repeat consultation. Patients are then asked to return if any problems or concerns arise.

Web-Deepening Procedures

Syndactyly Releases

For syndactyly releases in younger children, a well-padded plaster “sugar-tong” splint (see Fig. 47-19) or, in older patients, a palmar plaster splint, can be used. The hand is positioned in these splints with the wrist in neutral or slight extension and maintained for a period of approximately 1 week. The cast is then removed and the dressing is changed to avoid maceration. Subsequently, if the digits are short enough to be completely immobilized in a generous, well-padded, and adequately wrapped dressing, this is continued for another week or two with twice-weekly dressing changes. Otherwise it may be preferable to reapply a well-padded dressing and reimmobilize the hand in the same plaster sugar tong splint, a palmar plaster splint, or a thermoplastic splint for another week or two, again with twice-weekly dressing changes. After a total of 2 to 3 weeks of immobilization, a well-padded dressing should be changed regularly, keeping the hand protected (that is, in some children, this may mean continuing to keep the hand elevated and under the shirt), until healing is complete.

Multiple Z-plasties

Soft tissue surgery alone, without grafts, often benefits from a well-padded dressing for a total of 7 to 10 days. The dressing is usually changed after 1 week and the wounds are assessed. Afterward, daily dressing changes can be started. Immobilization in a plaster splint for comfort and to protect the incisions, especially in young children, may be useful in some cases.

Bone-Lengthening Techniques

Free Toe Phalanx Transfer

Bone-grafted toe phalanx transfer donor feet necessitate 4 to 6 weeks of K-wire immobilization of the operated toe^{71,89} in a non-weight-bearing posterior plaster splint, with the ankle immobilized in neutral position (that is, 90 degrees). Elevation of the foot to heart level reduces edema, especially in the first 2 or more weeks. Otherwise, if the donor defect is only closed by attaching the tendons and collateral ligaments together, a soft dressing and elastic bandage are applied, allowing the child to walk within 3 or 4 days,⁸⁷ keeping the foot elevated as much as possible the rest of the time. Cavallo et al,⁸⁶ who did not attempt to fill the donor defect, did not limit ambulation at all. For the hand, in these same patients, a well-padded plaster sugar-tong splint (see Fig. 47-19) can be used for immobilization. The K-wire and the cast are continued for 3 to 8 weeks total, depending on the author and details of the procedure.^{6,71,86,87,89}

On-Top-Plasty and Ray Transposition

In on-top-plasty and ray transposition in younger children, a well-padded plaster sugar-tong splint (see Fig. 47-19) can be similarly used for a period of approximately 3 to 4 weeks or more to allow consolidation of the bone and healing of any musculotendinous procedures. Thereafter, splinting may be required for variable amounts of time, depending on the procedure.

Distraction

Distraction With Immediate Bone Grafting

Length and type of immobilization depend on the method of fixation. For cases of distraction and immediate bone grafting immobilized with K-wires, the hand is placed in a short arm cast⁹⁸ or, in younger patients, in a well-padded plaster sugar tong splint, with the metacarpophalangeal

joints in moderate flexion and the wrist in slight flexion.⁹⁸ When the cast is changed, the wrist is brought into slight extension and the cast is worn for 5 to 6 weeks.⁹⁸ The K-wires are removed at 8 weeks, once consolidation is confirmed radiologically.⁹⁸ With the use of a plate and screws, immobilization can be shorter.

Gradual Distraction With Secondary Bone Grafting or Callotasis Bone Formation

In cases of gradual distraction lengthening, with or without secondary bone grafting, a soft dressing is applied, especially in small children,⁵ with or without a plaster splint,¹¹⁶ for the first week or two until the skin has healed. Subsequently some authors immobilize the hand in a plaster and/or thermoplastic splint for various lengths of time during distraction.^{94,99,116} Others have the patient begin range-of-motion exercises,⁹⁴ whereas still others encourage use of the hand in daily activities, even in cases of callotasis bone formation.⁹⁵ Daily pin care is recommended, beginning within a few days of the surgery.^{94,99}

The beginning of distraction is based on whether or not bone grafting is intended but mostly according to each surgeon's personal protocol. Some authors begin distraction within the first few days,^{5,6,97,99} whereas others begin between days 4 and 7,^{8,39,92,95} or even as late as 10 days postoperatively, with some maintaining compression at the osteotomy site before distraction.⁹⁴ If callotasis bone lengthening is intended, some suggest waiting for the appearance of some callus before beginning distraction.⁵ The rate of distraction also varies according to the author from 0.25 to 1.0 mm per day,* in increments of 2 to 4 times per day.†

During distraction, regular radiography and follow-up are initiated, anywhere from twice a week to once every 2 weeks,^{94,95} to ensure progress according to plan. The distraction rate is reduced if there is pain or extreme soft tissue tension.^{99,116} Adjustments in rate are also made according to bone appearance in cases of callotasis bone formation,^{94,95} often necessitating progression with smaller increments.¹²⁴ Distraction is continued until the desired length is achieved. Average distraction length varies from 25 to 70 mm in cases where the periosteum is transected and secondary bone grafting intended,^{5,6} and 10 to 30 mm or more when the periosteum is kept intact, allowing subsequent consolidation of bone callotasis to occur.^{8,88,93-97} The average time for distraction varies from 1 to 3 months.‡ In cases of callotasis bone formation, some authors describe bringing the patient to the operating room at the end of distraction if the skeletal position is not adequate to manipulate the bone regenerate and correct the apparatus position before allowing consolidation.⁹⁷

In cases of secondary bone grafting after distraction lengthening, immobilization is continued for 3 to 4 weeks if the stabilization of the graft is not rigid.⁹⁹ Some immobilize the hand in a splint and keep the K-wires for as long as 6 to 8 weeks when allograft metatarsal bone is used, and continue the splinting for another month after removal of the wires.¹¹⁶ Early mobilization is encouraged with rigid fixation of autografted bone.⁹⁹ In cases of callotasis bone formation, the distractor is worn for a total average time (that is, distraction plus consolidation) of 10.4 weeks to upward of 4 months, depending on the author.^{8,92-97}

Free Neurovascularized Toe Transfer

Dressing, immobilization, and other details of postoperative treatment are found in Chapter 51.

*References 5, 6, 8, 39, 88, 92-97, 99.

†References 5, 6, 8, 39, 92, 94, 95.

‡References 5, 6, 88, 92, 94, 97.

TREATMENT OUTCOMES AND COMPLICATIONS

In addition to the most common surgical complications that can occur intraoperatively (such as bleeding and damage to underlying structures) and postoperatively (such as problems with the dressing and infection), there are some complications particular to each of the various procedures previously described.

Web-Deepening Procedures

In web-deepening procedures, “shallowing” caused by scar contracture can necessitate repeat surgery. Skin graft loss after syndactyly release, although uncommon, may require repeat grafting, and often the need to switch to split-thickness skin grafts to improve the chance of “take.”

Bone-Lengthening Techniques

Free Toe Phalanx Transfer

In free toe phalanx transfer, if the chosen phalanx is either too wide or too long for the skin sleeve for which it was intended, the incision can dehiscence or skin necrosis can occur.^{5,6,71,87} Phalangeal displacement or instability can also take place.⁸⁶ Because the flexors have a stronger pull than the extensors, a flexion contracture can arise, particularly when the base of the phalanx is transferred to reconstruct a new metacarpophalangeal joint (J. Upton III, personal communication, 2006). Occupational therapy and splinting may help to correct the problem, which can be prevented by incising the tendinous “cap” so that the flexor component is somewhat longer in relation to the extensor component, as previously described. Damage to the epiphysis with subsequent fusion of the growth plate has been reported.⁸⁶ Resorption, with subsequent shortening, is particularly likely when the distal end of the toe phalanx has been removed during transfer.⁸⁶ If the surgery is undertaken after the child reaches 2 to 4 years of age, there may be little or no growth of the transferred phalanx,^{71,86,87,89} which also sometimes occurs in children who are operated on before 18 months of age.⁸⁶

The donor toe shortens if bone grafting of the donor site is not done, and can shorten even more if the flexor and extensor tendons are not sutured together to partially fill the donor defect.^{6,87} Shortening also occurs if there is damage or transection of the flexor tendon at surgery.⁸⁶ The toe remains unstable if bone grafting is not undertaken^{6,71} and may become displaced dorsally because of scar contracture of the dorsal incision or cross over an adjacent digit.⁷¹

On-Top-Plasty and Ray Transposition

In on-top-plasty and ray transposition procedures, the complications that are most likely to occur include neurovascular difficulties, problems with stabilization and correct positioning, and limited tendon function (that is, contracture⁶ or adhesions).

Distraction

In gradual distraction, problems with the distractor itself may occur. These can be mechanical in nature, including insufficient length of the apparatus to achieve the desired distraction,⁹⁵ breakage, wear and tear, poor upkeep, or even loss of the key.⁹⁹ Complications may also occur as a result of incorrect installation of the apparatus. Examples include the following: bending of the K-wires^{92,99} or fixation pins because of their inadequate size; placement of the fixation bar too close to the skin, thus causing injury⁹⁹; incorrect incision and positioning of the pins or wires with resultant tearing of the soft tissues⁹⁹; loosening of fixation into the bone⁹⁴ or tearing through it³²; extrusion,⁹⁹ angulation,^{94,99} or rotational deformity; and pseudarthrosis.⁹⁹ Even the palmar skin under the splint or dressing can be vulnerable.⁹⁹

Prolonged paresthesia of the digit^{88,98} is occasionally attributed to the use of the finger trap in one-stage distraction lengthening.⁹⁸ It can also be a result of excessive distraction of the neurovascular pedicle^{5,6} and may require slowing of the distraction rate.⁶ Pain, soft tissue damage,⁹⁹ fibrosis,⁹⁹ and bone exposure are possible if distraction is too rapid.⁹⁹ Bony prominences may also appear at the fingertips in cases of terminal bone distraction. These may be painful,^{94,96} necessitating stopping the distraction,⁹⁶ or performing other procedures such as trimming the bone or providing flap coverage.⁹⁴ Slowing of the distraction rate may be necessary with healing difficulties of the wounds^{5,6} or the skin bridge between the K-wires.⁵ Vascular compromise of the skin,^{5,6,39,88} pain,^{5,6,88} and progressive contracture may also require slowing of the distraction rate^{6,88} or stopping it altogether for a day or two.³⁹ Distraction of a more proximal bone, such as a metacarpal bone, can result in the progressive development of a flexion deformity at more distal joints, unless these are pinned in extension.^{5,6,88} There may be some loss of motion after distraction.^{95,96} Tendon adhesions may also be at fault in cases of distraction with secondary bone grafting, and these may require tenolysis.⁹⁹

An incorrect skin incision and placement of pins can result in maceration around the pins.⁹⁹ Development of a pin tract infection^{94,96} cannot always be avoided,⁸⁸ and can be caused by inadequate cleaning⁶ or nonrigid fixation of the device, as can osteomyelitis.⁹⁹ In the study by Dhalla et al,⁹⁴ all of the cases of infection occurred in patients treated with a single half-pin on either side of the osteotomy and longitudinal K-wire fixation during distraction; there were no infections in the double half-pin group. This may be caused by exposure of the intramedullary K-wire to the environment, thereby increasing the risk of infection. In their discussion, the authors suggest prophylactic antibiotic use in patients with single half-pins and a longitudinal K-wire distraction technique. They propose continuing the use of perioperative cephalexin for 2 weeks postoperatively and restarting it for another 2 weeks in the presence of pin track drainage or cellulitis. In some cases of deep infection, intravenous antibiotics may be necessary and the distraction device may have to be removed.⁹⁴ In patients with this complication who are undergoing callotasis bone lengthening, splinting may be sufficient to achieve consolidation, although secondary bone grafting may be required.^{8,94} Cold intolerance in patients living in cold climates is not unusual in children and is almost universal in adults.⁹⁹

Difficulty in achieving the planned length may result from fusion of the osteotomy site during distraction⁹⁷ or the use of the immediate bone-grafting technique. Final outcome can also be affected by bony angulation,^{88,94,96,99} pseudarthrosis,⁹⁹ or malposition.^{6,88,99} These sometimes occur when the rigidity of fixation is insufficient or when range-of-motion exercises are started too soon.⁹⁹ In cases of distraction with secondary bone grafting, malposition during distraction can sometimes be corrected at the time of the bone graft.⁶ Callotasis distraction lengthening may also occasionally require secondary bone grafting if callus formation is inadequate to achieve consolidation.⁹⁵ Fracture, during consolidation or after removal of the fixator, in callotasis lengthening⁹⁴ may complicate the final result. In cases of secondary bone allografting, sequestrum formation or fracture of the immobilization plate have been reported.¹¹⁶ Direct or vascular trauma to the epiphysis can affect the growth⁹⁹ of adjacent bones. In some cases, the lengthened and nonlengthened neighboring bones may not continue to grow in relative proportion to the rest of the hand, resulting in the worsening of initially acquired function such as pinch.⁹⁶ Finally, hypertrophic scarring⁹⁵ can downgrade even an excellent result.

Free Neurovascularized Toe Transfer

For toe-to-hand transfer, complications are detailed in Chapter 51. In addition to these, there can be problems associated with inadequate length, incorrect positioning, and limited motion of

the transfer. Limited motion of the transfer can be associated with poor quality of the proximal motors, which is most often seen in the more proximal forms of symbrachydactyly.⁷¹ Inadequate length and incorrect positioning are most often caused by poor initial planning. When multiple toes are transferred from the same foot, donor site defects can be disfiguring.¹⁰⁰ Adult patients with combined second and third toe transfers for congenital or traumatic injuries have more foot problems than those undergoing a single toe transfer.¹²⁰

Joint Stabilization Operations

As with any fusion technique, malunion and nonunion are potential pitfalls, especially when bone stock is deficient. It is important to treat soft tissues gently to avoid necrosis or healing difficulties of the soft tissue sleeves covering severely hypoplastic digits, or nail damage. Reconstructing ligaments to stabilize a joint while maintaining its mobility, when the joint surface area is inadequate, is aiming too high and will often result in failure requiring secondary fusion.

Other Corrective Procedures

Complications after other corrective procedures are similar to those found in normal hands undergoing the same procedure. It is nevertheless important to take into account that the structures are often smaller and/or hypoplastic, requiring adjustments in technique and reasonable expectations when planning surgery.

FUTURE DIAGNOSTIC AND TREATMENT MODALITIES

Improvement in the classification of this group of malformations would go a long way to gaining a better understanding of the genetics, incidence, associated malformations, and syndromes that can accompany several of the categories of undergrowth. It would facilitate the comparison of outcomes using various techniques of treatment with the aim of developing treatment algorithms according to the subclass and severity of a specific malformation. This would be particularly useful in clarifying the place of each of the many procedures described for the treatment of the various malformations presently classified as undergrowth. Furthermore, collaborative multicenter efforts would contribute to improving the care of all patients with hand malformations, particularly those with the rarer forms, as would well-maintained, complete diagnoses and complication registries.

Work in the genetics of hand malformations may eventually aid in the understanding and perhaps even in the treatment of certain hand malformations. Perhaps tissue engineering will provide eventual availability of “spare parts” for surgery. Although several hand transplants have been undertaken in adults, none has yet been performed in a child. Concerns about long-term immunosuppression and, in congenital hand cases, deficient recipient structures slow development in this area.

In the meantime, work is progressing in myoelectric prostheses, including improved motor control with the use of nerve transfers¹²⁵⁻¹²⁷ and research in the development of sensory feedback.¹²⁸ Most of the work is done in adult traumatic amputees, although some teams have been working with preschool and early school-aged children.¹²⁹ It is of note that, even if fitted at a young age, over half of children with absence of the segment at or distal to the wrist, will not use their prosthesis.¹²⁹ More recently, three-dimensional printers and devoted volunteers have given access to light, low-cost, colorful plastic mechanical hand prostheses that can be assembled by the patient and family and easily replaced as the child grows.¹³⁰

KEY POINTS

When to Operate

- Surgery should be reserved for patients with functional problems or when there is a clear functional benefit to intervening. It is paramount that the expectations of the surgeon, patient, and family be reasonable and realistic before embarking on a long and complicated reconstructive plan with all its potential risks and problems.
- Because patterns of hand function are generally established between the ages of 6 months and 2 years,¹⁰⁶ with thumb opposition beginning at approximately 7 months of age,¹⁰⁷ most operations modify the way the hand and the digits are used and thus are best performed before 2 years of age. Therefore, in chronologic order:
 - When a free phalanx transfer is appropriate, surgery should be undertaken under 1 or 2 years of age, and probably not after 4 years of age if one desires growth of the phalanx. For best results, the periosteum and ligaments should be kept intact.
 - Although amputation of tiny flail nubbins can be undertaken at any age, it may be wise to wait until it is clear that the nubbins are not useful for function and that a free toe phalanx transfer is either inappropriate or unwanted before proceeding. Similarly, ray amputation of a useless digit should not be undertaken before considering all potential uses for it. This should include development of a clear long-term plan that considers the possibility of transferring the digit or any of its components at a later stage during the process.
 - On-top-plasty and ray transposition are not commonly performed procedures; therefore there is not much in the literature about the ideal age at which these surgeries should be performed. Because these operations are mostly used to reconstruct the rays around the first web space, surgery should probably be performed early, but perhaps after the age of increased vasospasm. Between 1 and 2 years of age seems reasonable.
 - When possible, syndactyly release or multiple Z-plasties should be withheld until the patient can cooperate with postoperative care (after 3 years of age), unless there is a functional problem necessitating early intervention. In such a case, some operate on children at 6 months of age or younger, although waiting until they are 9 to 12 months of age facilitates surgery and decreases neurovascular risk as well as flap necrosis.
 - Ligament reconstruction, metacarpal rotation osteotomy, tendon transfers, and various other corrective interventions should be undertaken when the structures are large enough to allow stable, solid fixation. This usually means sometime during the preschool or early school years, be-

tween 4 and 6 years of age, or even later if the structures are particularly underdeveloped. Joint fusion should usually be delayed until the growth plates are closed to prevent secondary growth retardation resulting from trauma to the physes in bones that are already short. Some surgeons, nevertheless, may feel comfortable undertaking chondrodesis in younger children.

- In patients for whom distraction lengthening, with either immediate bone grafting, secondary bone grafting, or callotasis bone formation, is an appropriate modality, the age at which treatment is undertaken varies greatly in the literature. For this reason, surgery before school age should be considered in those patients in whom the potential functional benefits outweigh the risks. Otherwise, it is suggested to wait until there is good understanding and collaboration on the part of the patient, until larger bones permit more rigid fixation, and until there is less risk that disproportionate growth of the various elements of the hand will undo the correction—perhaps even as late as adolescence.

Interdisciplinary Approach

- Because of the complexity in treating some cases of undergrowth, it is suggested to have a well-integrated interdisciplinary approach for some patients:
 - Pediatrics and/or medical genetics can screen for syndromic cases and refer the patient to other relevant specialists or multidisciplinary clinics.
 - Occupational therapy and/or physiotherapy can evaluate function, help plan treatment, undertake postoperative therapy, and follow the patient for deterioration with growth.
 - Psychologists and/or social workers can assess the patient's and family's ability to cope with the malformation and multiple proposed interventions, as well as support the patient during various stages of treatment and life's milestones.

The Treatment Plan

- Very mild cases of undergrowth require no treatment at all.
- More significant digital shortening may benefit from syndactyly releases to improve handbreadth, but at the expense of scars, which may or may not be considered a fair exchange in the eyes of some families.
- Various reconstructive procedures such as first metacarpal rotational osteotomies, tendon transfers, ligament reconstructions, joint fusions, on-top-plasties, ray transpositions, use of nonfunctioning bone segments as grafts, and so forth, must be considered and planned at the onset, even if the actual surgery is not going to be undertaken until much later.

Continued

KEY POINTS (continued)

The Treatment Plan—cont'd

- Because phalangeal lengthening does not permit the creation of new joints, which results when they are elongated excessively in stiff, thin digits, distraction is more often used for the metacarpals. When used for phalanges, the technique is probably most appropriate when there is not only significant shortening but also an associated deformity, such as angulation, that requires correction. It may be useful to consider distraction of an otherwise useless segment with the aim of transposing it as an on-top-plasty, usually on either side of the first web. Distraction is also useful for the reconstruction of pinch, including its use for some ray transfers.
- The choice of a distraction technique depends not only on the preference of the surgeon, the patient, and the family, but also on the amount of lengthening required. Distraction lengthening with immediate bone grafting permits approximately 5 to 15 mm of lengthening. Distraction lengthening with callotasis bone formation achieves approximately 10 to 30 mm of additional length. Finally, distraction lengthening with secondary bone grafting permits approximately 25 to 70 mm of lengthening.
- A free toe phalanx transfer can be considered in young patients (less than 1 or 2 years of age, and rarely after 4 years of age) to stabilize flail skin sleeves. The result is superior when the metacarpophalangeal joint is present and actively mobile.
- Amputation should be reserved for tiny, flail nubbins that are not useful functionally and cannot benefit from a free toe phalanx transfer. Ray amputation is indicated for a flail, single hypoplastic digit in a normal or near-normal hand.
- Multiple Z-plasties permit deepening of the web that is present between a remaining thumb and the little-finger ray in patients with oligodactylic/apthalangic symbrachydactyly. This procedure widens the breadth of the grip, allowing the patient to grasp larger objects without losing fine pinch. The intervening hypoplastic metacarpals should not be further shortened before making sure that they are not useful in stabilizing grip.
- A free neurovascularized toe transfer is recommended for more severe cases requiring reconstruction of some form of pinch. The aim is to achieve a two- or three-digit pinch, including any available digits in the hand that can be reconstructed or stabilized. It is important to note that the more proximal the deficit, the more severe the malformation and the less likely that adequate proximal motor function is present. This can affect the final range of motion and end result. Anticipated range of motion must be taken into consideration when planning the surgery, including positioning and adjustment of the transfer.

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49

Amniotic Band Syndrome

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central feature of *amniotic band syndrome* (also known as *amniotic band sequence*, *constriction ring syndrome*, *congenital constricting bands*, and *congenital ring syndrome*) is the presence of asymmetrical, nonanatomically distributed, soft tissue abnormalities of the extremities. Extremity malformations can be classified into five major groups, first proposed by Patterson¹ (Fig. 49-1). Anomalies that can occur in association with amniotic band syndrome include clubfeet, facial clefts, cleft lip and palate,² body wall defects,³ and cutis aplasia. The most severe cases may lead to anencephaly or fetal demise.⁴ Most cases are sporadic. The estimated incidence is approximately 1 in 10,000 live births, with an equal sex distribution.^{1,5} No genetic associations have been found,^{1,6} and more than half of documented cases in one report had abnormalities during gestation, including prematurity, maternal illness, low birth weight, and maternal drug exposure.⁷

GENETICS AND CAUSE

Numerous observers since the time of Hippocrates have described newborn infants with apparent amniotic band syndrome. Van Helmont of the early seventeenth century was one of the first to describe an intrauterine amputation.⁸ Montgomery,⁹ in 1832, described free strands of tissue located at the site of a constriction band. Intrinsic and extrinsic mechanisms have been proposed.

In 1930 Streeter¹⁰ stated that the cause of the constriction ring syndrome was intrinsic to the fetus. Inadequate fetal “germ plasma” leads to a local degeneration of soft tissue in a manner analogous to a cleft lip and palate. The intrinsic theory survives, because of a higher-than-expected association of this syndrome with internal malformations such as absent or dysplastic kidneys, ectopically placed gallbladders, imperforate anus, and loss of the abdominal wall. The intrinsic theory is supported by cases of amniotic band syndrome in twin gestations. In most twin gestation cases, both fetuses are affected, suggesting a phenomenon that is systemic (genes, maternal factors, or teratogens) rather than local. Streeter hypothesized that the fibers seen at the bases

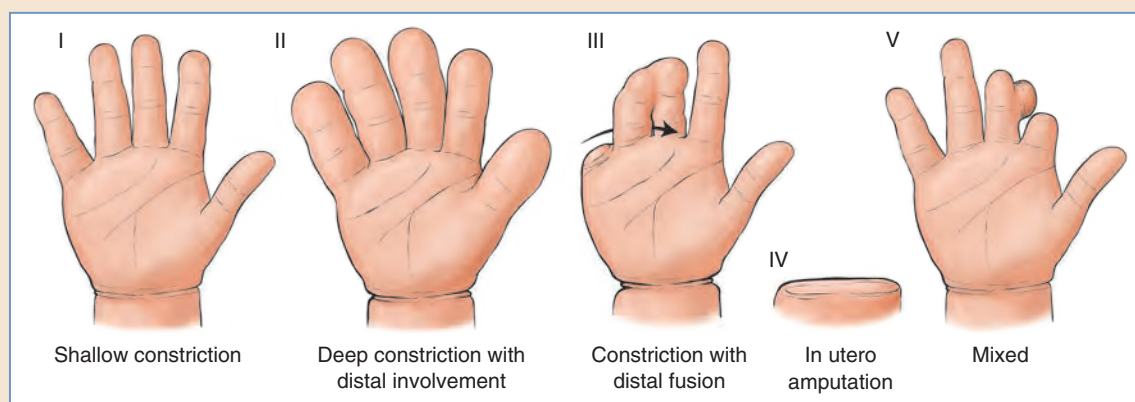


Fig. 49-1 The major consequences of amniotic bands, modified from the Patterson classification.¹ (I, Shallow constriction; II, deep constriction with distal involvement $\pm$ lymphedema; III, constriction with distal fusion (acrosyndactyly); IV, in utero amputation; V, mixed [can include fusion of the amputated part to a different finger].)

of bands were attempts at fetal wound healing. Finally, breaches of the amnion (a cornerstone of the extrinsic theory) occur with each successful amniocentesis performed, yet the incidence of amniotic band syndrome does not increase after this procedure.¹¹

The favored theory postulates an extrinsic pressure applied to the tissues of the growing fetus. The extrinsic theory, particularly its description of pressure and necrosis of tissue, makes intuitive sense to those in surgical fields. Torpin¹² hypothesized that amniotic band syndrome is caused by an in utero tear in the placental amnion. The fetal skin is in continuity with the amnion at the umbilicus, and deep to the amnion is the chorion. When the amnion ruptures spontaneously early in the middle trimester, it can separate from the chorion. This produces tiny strands of free-floating tissue. The fetal hands and feet, because of their waving motion, “catch” these 1 to 2 cm long strands. If the strands were to embed into the fetal skin, further growth of the rapidly enlarging fetal extremity would be limited, resulting in a constriction ring of variable severity. Rarely, these strands can be recovered from the base of a newly born infant with amniotic band syndrome. In one case, Kiehn et al¹³ released a fibrous band of inelastic material from the leg of a twin born at 27 weeks with an underlying constriction ring. Mechanisms by which these strands can cause various types of anatomic changes in fetal limbs are easily postulated.

Rupture of the amnion causes a temporary oligohydramnios because of exposure of the amniotic fluid to the relatively permeable chorion. Oligohydramnios persists until the chorion thickens (Fig. 49-2). Oligohydramnios and increased intrauterine pressure on the fetus can explain the coincidence of clubfoot and cleft palate (akin to Pierre Robin sequence) with the amniotic band syndrome.¹⁴ A recent report of iatrogenic pressure on a growing fetal limb as a cause of a constriction band supports the extrinsic hypothesis.¹⁵

In utero amputations provide a clue about the timing of the amnion rupture and the onset of pressure on the developing limb. This was documented in several cases in which the amputated

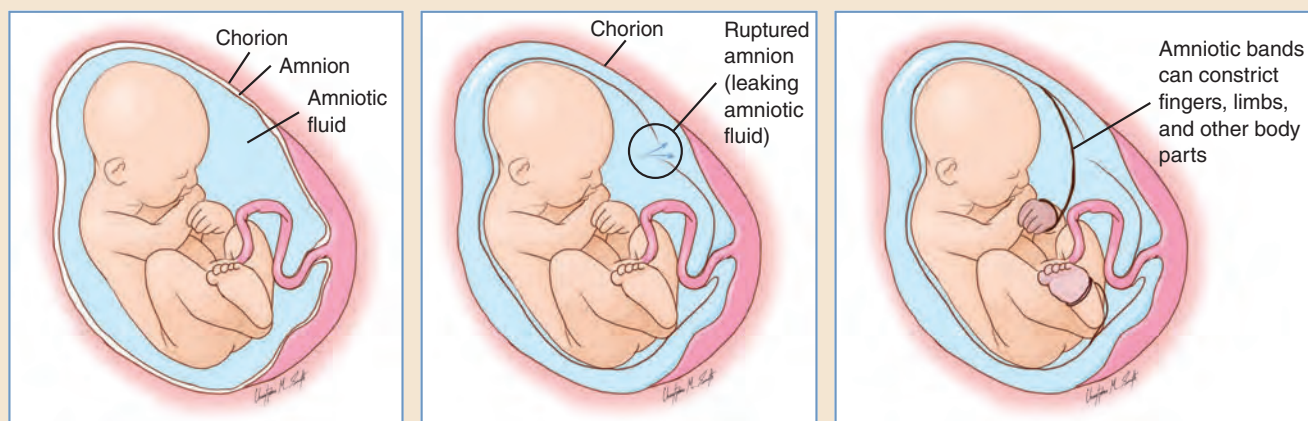


Fig. 49-2 Chorioamniotic separation.

piece was located after the birth of the child. In most of these cases, the amputated limb had a gestational size of older than 14 weeks and younger than 20 weeks.

A fetus can swallow large amniotic cords. This draws the vertex of the fetus toward one site on the placenta.¹⁶ The direct contact, restricted motion, and constant pressure of the fetal head on the placenta may be a cause of the cutis aplasia sometimes seen with amniotic band syndrome (akin to a fetal pressure sore). Nonanatomic perioral clefts are associated with the syndrome and hypothesized to be caused by the indentation of a swallowed cord on perioral tissues.¹⁷

DIAGNOSIS

Prenatal

Prenatal diagnoses with fetal ultrasound and fetal MRI facilitates prebirth counseling and preoperative planning. For the past 20 years, ultrasound has been useful for identifying swollen limbs and areas of constriction. Three-dimensional ultrasound helps parents to appreciate the deformity and surgeons to start planning and having discussions with the parents prenatally.¹⁸ Some constricted areas seem to resolve spontaneously; therefore serial ultrasound examinations of the fetus are important.¹⁹ One report documented decreases in blood flow distal to the constriction band, which identifies limbs at risk for in utero amputation.²⁰ For limbs at risk, fetoscopic release of bands has proved feasible in selected centers, again necessitating a balance between the health of the fetus and the mother and the survival of the limb.²¹ In a review of seven cases of fetoscopic release of amniotic bands, Hüsler et al²² found premature rupture of membranes in 71% of patients, delivery at a median of 34.8 weeks, and one case of intrauterine fetal demise. One case without limb perfusion at the time of fetoscopic release eventually required amputation after birth, and another case with failed fetoscopic release nonetheless had a good outcome with surgery after birth. The authors concluded that the cases likely to improve had abnormal but present blood flow distal to the constriction band.

Postnatal

The hallmark of amniotic band syndrome is an inelastic transverse extremity band of variable width and depth. Tissues proximal to the ring typically appear normal. The patterns of extremity involvement are varied—a single extremity can be affected at several levels, and each patient often has multiple extremities affected. The bands can be shallow or deep and result in distal fusion or in utero amputation. The middle and ring fingers are the most commonly affected.²³ The thumb is somewhat spared because of its short length and relatively protected abducted volar position in utero. Affected digits have a high percentage of fingernail hypoplasia or absence (Fig. 49-3).

Differential Diagnosis

The diagnosis of amniotic band syndrome is relatively straightforward for cases of deep bands with concomitant distal deformity (such as lymphedema). The diagnosis is less straightforward for infants born with syndactyly, acrosyndactyly, or apparent in utero amputations, because these conditions can occur in isolation. To make the diagnosis for children with amputations or acrosyndactyly, another finding such as involvement of an additional extremity, cutis aplasia, clubfoot, facial clefts, or abdominal wall defects is needed. Other distinguishing features on a physical examination of the infant indicate this syndrome. For amputations, bare bone of apparent normal size may be found protruding past the level of the soft tissue amputation, as if a guillotine amputation had been performed and the soft tissues had retracted. This is in contradistinction to cases of transverse arrest, in which atretic soft tissues cover a hypoplastic bone. Amniotic band syndrome patients with acrosyndactyly have the unusual physical finding of epithelialized sinuses at the bases of the fused digits. Finally, adjacent to an amputated digit, the adjacent piece of soft tissue often appears enlarged, as if a portion of the amputated digit has survived in part as a soft tissue graft. These subtle findings from a physical examination suggest a slow ischemic insult to the tissue distal to the constriction band.

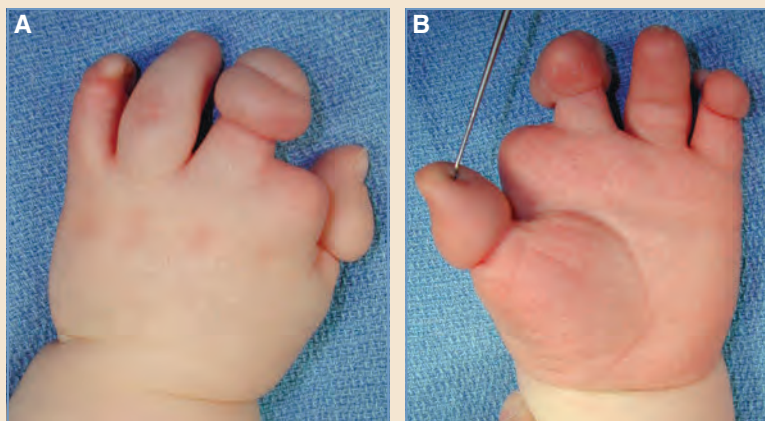


Fig. 49-3 The left hand of a 10-month-old infant with amniotic band syndrome. The index finger suffered an amputation in utero, and a portion of the digit survived as a graft onto the dorsum of the middle finger. **A**, Anterior view. **B**, Posterior view.

CLASSIFICATION, TIMING, AND SURGICAL MANAGEMENT

The natural history of minor constrictions is variable. Patterson¹ thought the bands were most noticeable at birth, because the indentations are elevated in high relief by an infant's baby fat. In contrast, Flatt²³ wrote that in more than 50% of the infants, the constriction worsens over time because of cicatricial healing of granulation tissue at the base of the bands. For this reason, children with shallow constriction rings should be followed closely after birth. In a case report, Ho et al²⁴ treated a premature, critically ill infant born with a well-vascularized soft limb that progressed to dusky and tense on day 4 of life. The child required an emergent simple band release at the bedside, and the edema resolved. Samra et al²⁵ and Weinzwieg²⁶ also presented cases of rapid limb swelling and vascular compromise after birth that improved with immediate surgical intervention.

We propose a modification of Patterson's classification system, with some insight from Weinzwieg²⁶ and Hall et al,²⁷ who noted, respectively, the importance of distal lymphedema and evolving vascular compromise, and we add a focus on neuronal involvement.¹ Our system attempts to base treatment on anticipated limb function and to aid in determining the best timing and type of surgical management (Fig. 49-4). This classification ranks the deformities from the most to the least urgent. Deep constriction bands with vascular or nerve involvement are urgently

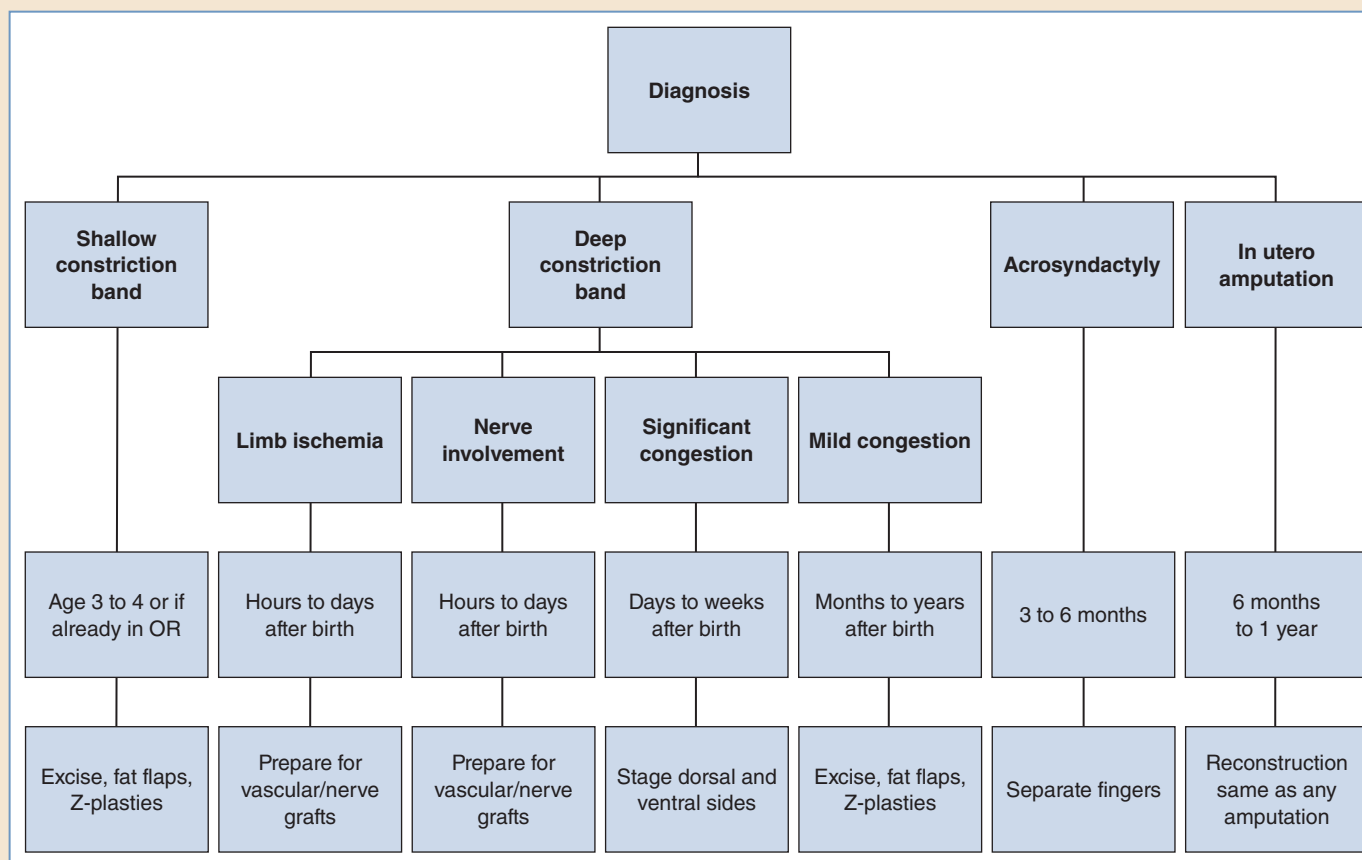


Fig. 49-4 Amniotic band syndrome classification and treatment algorithm.

addressed, whereas less urgent deformities, including deep constriction bands with venous or lymphatic congestion and acrosyndactyly, can be treated within days to months after birth. In utero amputations do not require urgent treatment and, if indicated, can be reconstructed when patients are 6 months to 1 year of age. Finally, shallow constrictions are treated as an elective procedure based on the degree of cosmetic deformity.

Shallow Constriction Bands

The indication for treating stable, shallow constriction rings is cosmesis. The severity of a cosmetic deformity should be balanced against the risk of anesthesia when deciding the optimal timing for revision. As a child ages, the neurovascular structures become larger and easier to dissect. Though it is a matter of opinion and clinical practice, shallow bands are generally not treated in our center before patients are 3 to 4 years of age unless they are in the operating room for another reason.

Similar to the surgical procedure described by Upton and Tan,²⁸ the treatment of shallow constriction bands includes loop magnification and excision rather than incision of the band (Fig. 49-5). After skin flap elevation, subcutaneous fat flaps are developed, and the dorsal aspect of a prominent fat flap can be debulked. The fat flap is mobilized across the band for improved contour, and a straight dorsal closure is performed. Finally, Z-plasties are performed on the lateral aspect of the digit or extremity (Fig. 49-6). Shallow bands may be completely revised in one procedure, even if the band is circumferential, and results tend to be quite good.^{28,29}

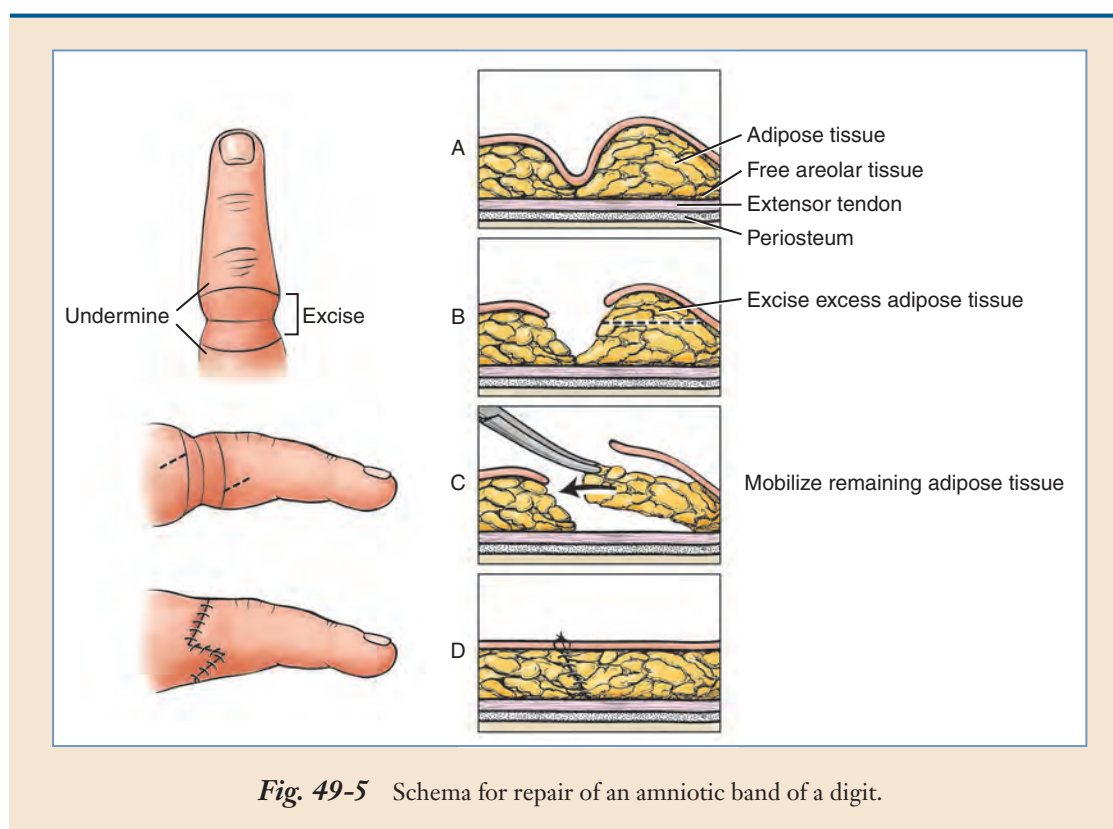


Fig. 49-5 Schema for repair of an amniotic band of a digit.

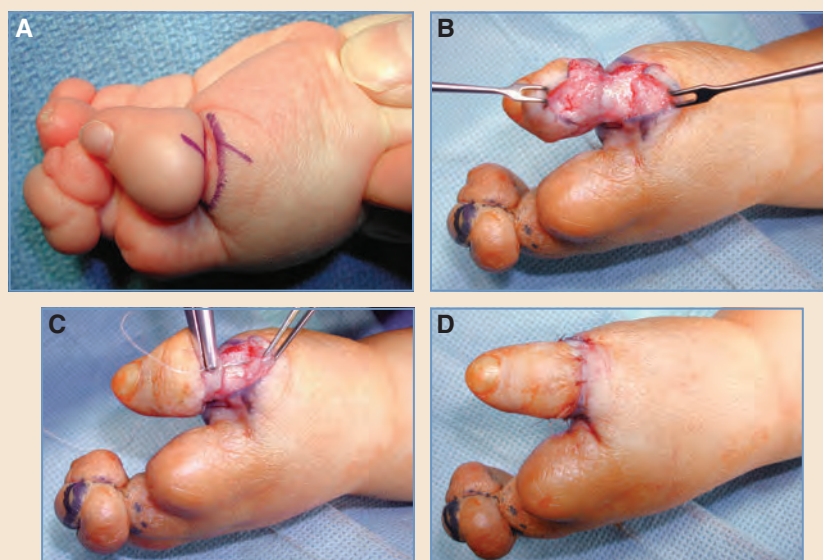


Fig. 49-6 The thumb of the hand shown in Fig. 49-3, treated for an amniotic band. **A**, Excision of ring and midlateral Z-plasties are marked. **B**, Skin flaps are retracted to contour the dorsum of the fat of the digit. **C**, Fat flaps are mobilized across the band. **D**, The thumb is shown after skin closure.

Although early methods of amniotic band reconstructions of the limbs have included circumferential Z-plasties, more recent articles have proposed modifications including turnover dermatofascial flaps with rectanguloplasty instead of Z-plasty to orient the scars along relaxed skin tension lines.³⁰ Another article proposed straight-line closure to further decrease scar burden.³¹ The authors thought it reasonable to perform a straight-line closure with two posterolateral Z-plasties in the limb (as shown in Figs. 49-5 and 49-6) to combine the advantage of a straight-line scar with the minimization of scar contracture by the Z-plasties in areas that are less visible. The unifying theme of all techniques is complete excision of all segments of the amniotic band.

Deep Constriction Bands

Most surgeons generally agree that staged treatment of deep constriction bands is safer than a one-stage circumferential revision. Staged treatment is thought to lessen the chance of distal circulatory compromise caused by perioperative swelling, tissue trauma, and interruption of subdermal plexus circulation. The second stage is performed 2 to 3 months after the first procedure. Surgical principles are similar to those followed for treating more shallow bands. Great care is taken to preserve all veins and lymphatics that run longitudinally across the band. An operating microscope is useful for this.

A one-stage repair of deep circumferential bands with distal involvement is performed in some centers. They correctly think that it prevents a second trip to the operating room^{27,32} and may reduce possible digital compromise from postoperative swelling and potentially tight dressings. They argue that, because of the deep bands, venous and lymphatic flow are already channeled along the tendons of the finger and along the fascia of the extremity and therefore would

not be divided by a circumferential skin incision. The location of the arteries is predictable, and these can be protected during the dissection.

Limb Ischemia

The most urgent and most difficult deformities are related to deep constriction bands. For such bands with documented limb ischemia, we recommend surgery within hours to days of birth. The discussion with the family needs to include the possible need for vascular and nerve grafts and the possibility of long-term growth restrictions and permanent nerve deficits if the release is not performed. We recommend reconstructing underlying major nerves and vessels with grafts at the initial surgery. In one study of 8 patients with a total of 20 affected limbs, 100% of the limbs with deep bands or with amputations were found to have anomalies or absence of major arteries proximal to the band on MRA or CTA, whereas superficial bands had only an 11% incidence of an underlying vascular anomaly.³³ Weinzwieg²⁶ concluded that this may be secondary to distal maldevelopment and thus attrition of the blood supply or lack of formation of the normal vascular pattern. Weinzwieg²⁶ showed good short-term outcomes with immediate decompression to evacuate lymphedema fluid, staged band excision, Z-plasty, and local wound care for distal tissue necrosis. Samra et al²⁵ presented a 6-year follow-up of a significantly compromised limb that was salvaged by immediate excision of two thirds of the band, followed by release of the remaining third a week later and sural nerve grafting to the sciatic nerve at 6 months for lack of motor and sensory function. The 6-year outcome included a 5.5 cm leg length discrepancy and decreased sensation on the dorsum of the foot; however, all other sensation and motor function had returned, and the patient was able to run, ride a bike, and balance on each foot.

Nerve Involvement

Nerve involvement caused by compression and/or ischemia with associated sensory or motor dysfunction at birth are additional severe manifestations of amniotic band syndrome. Surgery with nerve grafts is recommended within hours to days of birth. In an interesting subset of patients, nerve morphology distal to the constriction appears normal, suggesting that the ring occurred after the formation of the nerve trunks. In several instances, the intraoperative appearance of the nerve did not correspond well to the preoperative assessment of nerve function clinically or by nerve studies.^{34,35} Despite early (3 to 6 months) exploration and intrafascicular release, long-term return of function has not been exceptional. This has led some surgeons to advocate even earlier surgical releases of the involved site and being ready to excise and graft the nerve across the constriction site.³⁶

Lymphatic or Venous Congestion

Less severe constriction bands only affect the low-pressure venous and lymphatic systems. The distal edema is thought to be of lymphatic origin. Patients with deep rings often have temperature differences of several degrees centigrade between affected and unaffected digits. Typically, distal affected skin becomes violaceous after cold exposure. Early release of the band reduces the pressure gradient that must be overcome for lymphatic return. The more congested the distal extremity, the earlier the revision of the band must be performed. For significant congestion, surgery is recommended within days to weeks to prevent long-term sequelae.

In a physical examination, generalized swelling distal to a constriction ring should be distinguished from a localized, nonswollen mass distal to a band. For patients with a localized, non-



Fig. 49-7 The right hand of the same 10-month-old infant (see Figs. 49-3 and 49-6) with acrosyndactyly. A snap passes atraumatically through the base of the web space.

swollen mass distal to a band, if an adjacent digit had an in utero amputation, the mass may be the remnants of the amputated digit that survived by grafting. Therefore this digit with a localized mass can be treated more like a digit with a shallow constriction band rather than urgently, like an extremity with a deep constriction ring.

Acrosyndactyly

Acrosyndactyly is from the Greek *akros*, meaning “topmost.” It is constriction with distal fusion of soft rather than bony tissue, with more distal versus proximal involvement. Patterson¹ has further subdivided acrosyndactyly into the following:

- I Fingertip fusion with webs intact
- II Fingertip fusion with partial webs
- III Fingertip fusion with sinus tracts, but no webs

Most cases have at least some degree of web space shortening. When multiple digits are involved, the digital tips are said to resemble a small bunch of grapes, and assigning a fingertip to its appropriate digit is quite difficult (Fig. 49-7). Surgical release should be performed by 3 to 6 months to maximize the chance for proper longitudinal bone growth and to allow the child to use the fingers independently.^{37,38}

Surgery for these patients follows the principles used to treat an Apert hand. Full-thickness skin grafts are used liberally; web spaces are reconstructed with broad flaps (the skin of the sinuses is not overly broad or helpful); digits are resected when appropriate; and surgery should be restricted to one side of a digit at a time to prevent vascular compromise.

When all five digits are involved, we typically follow a sequence to release the first and third web spaces at the initial procedure, followed by a release of the second and fourth spaces. This allows a two-stage release of all digits without the need to perform a procedure on both sides of the same digit. Further deepening of the web spaces by release of the deep transverse metacarpal ligament with or without intrafascicular nerve dissection may also be necessary to lengthen the short digits in cases of severe acrosyndactyly.

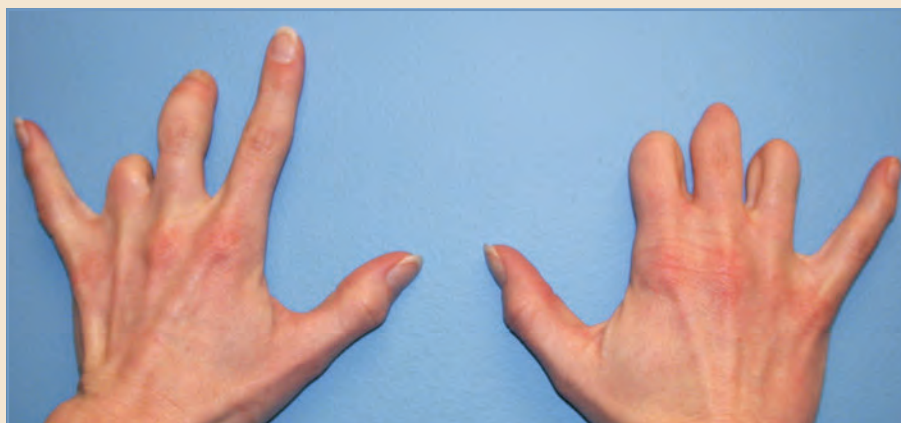


Fig. 49-8 This untreated adult with amniotic band syndrome has no complaints regarding hand function.

In Utero Amputations

Little is written concerning the management of children with in utero amputations, possibly because the indications for active intervention are few. In comparison to cases of transverse arrest, patients with amputations from amniotic band syndrome do not have associated soft tissue digit nubbins; therefore they do not need revisions for cosmesis or function. Adults with loss of multiple digits by birth often have near-normal hand function (Fig. 49-8). Patients with unilateral involvement typically report that they use the affected limb as a helper, and involved reconstructions or prostheses are typically not necessary or indicated.

Infants with bilateral digital amputations may be helped by skeletal distraction for digital lengthening to improve grasp.³⁸ Alternatively, a resected second metacarpal can be used as a spare part to augment a congenitally amputated thumb.³⁹ This procedure simultaneously increases thumb length while it deepens the first web space. Finally, microvascular toe transfers are possible, because the toe tendons can be anastomosed to normal proximal tendons in the palm and on the dorsum of the hand. Surgeons can consider an “on-top” plasty, toe-to-thumb transfer, or web-space deepening as appropriate in patients 6 months to 1 year of age.

OUTCOMES

Long-term outcomes of surgery for amniotic band syndrome are mixed. The outcomes for shallow bands are uniformly good. However, growth disturbance seems to be increased with greater depth of the band and associated vascular compromise. One of my (G.A.D.) patients underwent a release of an upper arm constriction band, neurolysis of the median and ulnar nerves, and a brachial artery exploration at 7 months of age and continues to have arm length discrepancy and impaired function at her 6-year follow-up (Fig. 49-9).

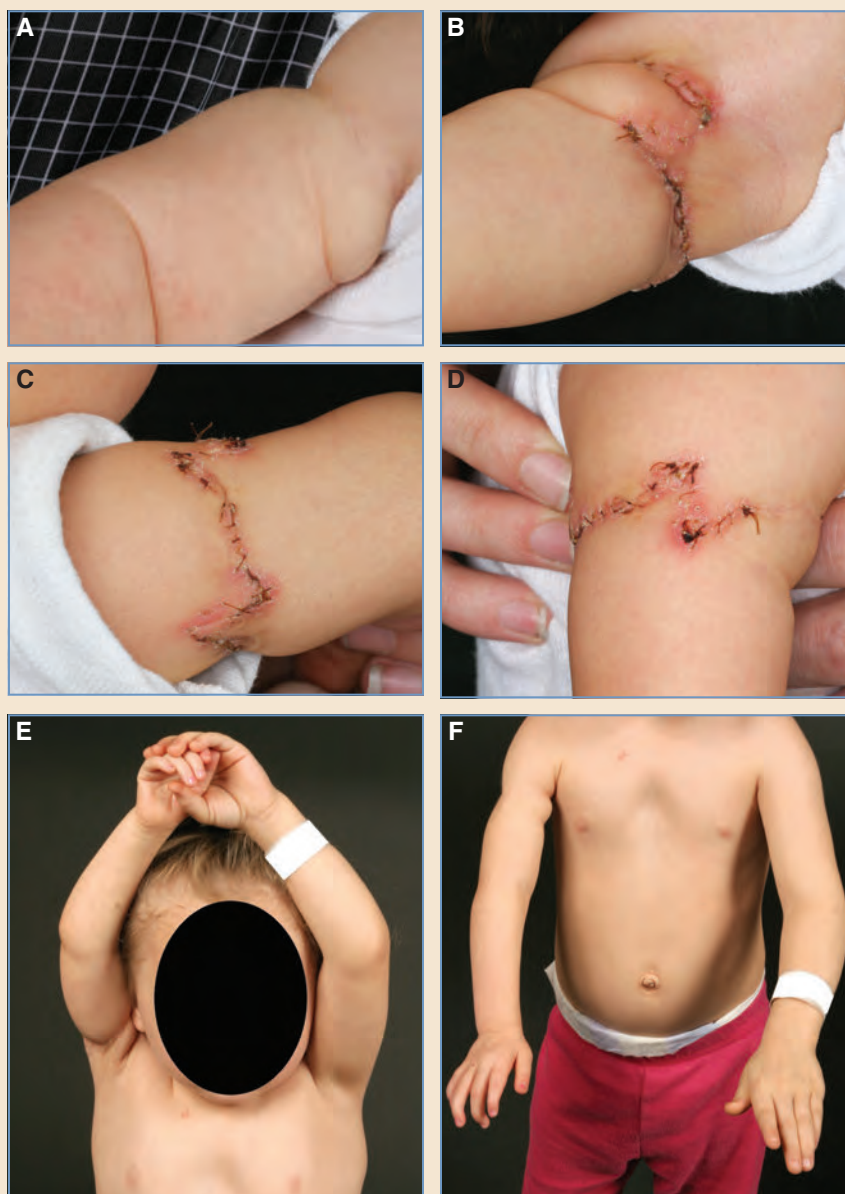


Fig. 49-9 A, This 7-month-old female had a right upper arm constriction band, decreased arm growth, and a cool arm. B-D, Medial, lateral, and dorsal views show the patient at 8 months of age, after exploration of her right brachial plexus, multiple Z-plasty closure of the constriction band, neurolysis of the median and ulnar nerves, and exploration of the brachial artery. E and F, Two years postoperatively, the patient uses her left arm primarily and has decreased growth of her right upper extremity and no independent finger extension and flexion. She can make a composite grasp in extension and has good biceps and triceps function.

CONCLUSION

Advances in our understanding of amniotic band syndrome will continue to evolve. We will be at the forefront of these advances. Surgery has a wide range of applications from pure cosmesis to functional improvement and limb preservation.

KEY POINTS

- All patients with constriction bands should be examined early by a surgeon and followed closely for congestion and nerve or vascular changes during the first few weeks to months of life.
- Bands with nerve or vascular compromise should be treated aggressively and may need nerve or vessel grafts for reconstruction in the neonatal period.
- Deep bands should be released in a timely manner to prevent permanent swelling and dysfunction of the distal segment.
- Complete one-stage circumferential treatment should be avoided because of the risk of venous and lymphatic congestion.
- Patients with acrosyndactyly are analogous to patients with Apert syndrome, requiring large skin grafts for release of digits.
- Patients with full amputations often have excellent function and usually do not require surgical treatment.
- Shallow bands are cosmetic issues. The risks of treatment should be balanced against the potential benefits.
- Three-dimensional ultrasound has increased the accuracy of prenatal diagnosis and allows early family discussion and surgical planning; however, in utero fetoscopic band lysis, although successful in a few cases, carries significant risks of morbidity and mortality for a disease that is not life-threatening.

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Generalized Systemic Conditions That Affect the Hand

Reuben A. Bueno, Jr. • Ronald M. Zuker • Benjamin Alman



Generalized systemic conditions can affect both the appearance and function of upper extremities. Regardless of whether the pediatric patient with a hand deformity or hand problem is seen with or without a known diagnosis, the consultant hand surgeon should be aware of the generalized systemic conditions that may affect the hand and upper extremity. In some presentations, the surgeon may be the first specialist to evaluate the patient and make a diagnosis. Although the presentations may vary, depending on the underlying disease, the common goal is optimizing the function of the hand and upper extremity. Surgery is only one aspect of a multidisciplinary approach for a patient with a systemic disease. As such, careful planning with realistic expectations is necessary when considering surgery to improve function and appearance. Treatment can be challenging—functional improvement may be temporary, because the propensity for recurrence is high in patients with systemic conditions.

MULTIDISCIPLINARY APPROACH FOR A PATIENT WITH A CHRONIC SYSTEMIC CONDITION

The chronic nature of many systemic diseases requires recognition by the hand surgeon that surgery is only one component in the total care of the patient. In addition to the general care provided by the pediatrician or family doctor, other specialists may need to be involved; for example, a neurologist for neuromuscular conditions, a rheumatologist for juvenile rheumatoid arthritis, a dermatologist for epidermolysis bullosa, or a geneticist for various contracture conditions.

Equally important to the management plan are therapists and nurses, who may be in contact with these patients on a more regular basis than physicians. If surgery is to be considered, input from all members of the multidisciplinary team should be sought to make the best recommendation for the patient.

GENERALIZED SYSTEMIC CONDITIONS THAT AFFECT THE HAND AND UPPER EXTREMITY

Many systemic conditions may affect the hand, and in some patients, the entire extremity. An adequate functional assessment of the hand must take into account the position and function of all the joints of the upper extremity, including the shoulder, elbow, and wrist. This chapter will address those conditions most likely to be seen by the hand surgeon, with consideration given to how these conditions affect the other joints of the upper extremity. These conditions include the following:

- Skin disorders: Epidermolysis bullosa (EB)
- Contracture disorders: Arthrogryposis
- Neuromuscular disorders: Cerebral palsy (CP)
- Inflammatory disorders: Juvenile rheumatoid arthritis (JRA)
- Hematologic disorders: Fanconi anemia (FA)

Skin Disorders: Epidermolysis Bullosa

Epidermolysis bullosa is a generalized systemic condition characterized by blistering, ulcerations, and scarring of the skin after minor trauma. The four major groups of EB are (1) EB simplex, (2) junctional EB, (3) dominant dystrophic EB, and (4) recessive dystrophic EB (RDEB). More than 20 subtypes of EB have been described.¹ Patients with EB have fragile skin; hands are especially susceptible to blistering and scarring because of the repetitive trauma of daily activities (Fig. 50-1). Patients with RDEB have the most deformed hands; patients with the Hallopeau-Siemens subtype are the most severely affected.²

Repeated trauma to the skin of the hand and subsequent scarring can lead to progressive deformity, ranging from flexion contractures at the interphalangeal (IP) joints (Fig. 50-2) to the characteristic *pseudosyndactyly*, which is caused by fusion of the web spaces, to “mitten” or “co-



Fig. 50-1 A, Dorsal and B, palmar views of the hands of a patient with EB. The thin, fragile skin is susceptible to recurrent blistering, ulcerations, and scarring from minor trauma.

coon” deformity (Fig. 50-3). This restrictive cocoon is composed of a hyperplastic epidermis, with a stratum corneum five times as thick as the remaining epidermis.³ Soft tissue scarring with the fingers and wrist in a flexed position can eventually cause joint contractures and bony erosion. Intrinsic hand muscles may also atrophy because of lack of use. There is variability in severity of skin and joint involvement in EB patients at presentation. Scarring at an earlier age has been associated with more severe deformities later in life.²⁻⁷ Hand deformities are seen in 95% to 98% of EB patients with the Hallopeau-Siemens subtype and in approximately 40% to 50% of patients with all other types of RDEB.²

Once the deformity affects hand function, nonsurgical options such as therapy and splinting are not effective to improve function or prevent worsening of the deformity. Because splints can result in repetitive trauma and scarring, they should be used judiciously. When considering surgery, all members of the multidisciplinary team must evaluate the patient’s overall health and ability to tolerate surgery. The quality of the skin, the patient’s nutritional status, and the involvement of other organ systems are assessed. The quality and amount of available skin are assessed, so that coverage with adjacent skin, a skin graft, or skin substitute can be planned. As the skin contracts and leads to worsening contractures, the joints of the digits and the wrist can become secondarily involved. Radiographs should be obtained to check for evidence of arthritic change and bony deformity. Surgical options will be affected by the presence of bony involvement. Evaluations by physical and occupational therapists should measure both the range of motion at affected joints and the patient’s ability to perform activities of daily living. Postoperative care includes immobilization, followed by splinting and therapy. Whether a patient can tolerate the



Fig. 50-2 Proximal interphalangeal (PIP) joint contractures in a patient with EB resulting from recurrent soft tissue scarring. Note the pseudosyndactyly from fusion of the web space between the ring and small fingers, as well as the loss of nail anatomy and shortening of the digits.

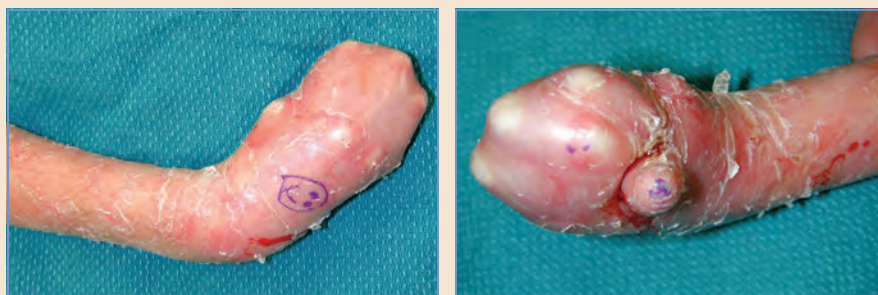


Fig. 50-3 Mitten deformity in a patient with EB.

postoperative regimen should be discussed before surgery, and input from all who provide care to the patient should be considered.

Because of the chronic nature of EB and the resulting progressive worsening of the hand, surgery should be viewed as a salvage procedure to preserve as much function as possible. Patients with the Hallopeau-Siemens subtype are the most likely to undergo surgery to improve hand function; these patients average three procedures per hand.² Unfortunately, there is no definable endpoint for hand surgery in an EB patient. As with other chronic conditions, the surgical plan must be individualized, taking into account the patient's overall health and level of function. During the preoperative period, nutritional status should be optimized with supplements and possibly a feeding tube. Hemoglobin levels should be measured; epoetin alfa (Epogen) has been used to prepare the patient for surgery. Hematologic consultation is recommended if Epogen is to be considered. Potential donor sites for skin grafts are evaluated, or arrangements are made for skin substitutes to be available for wound coverage. Because intubation may be complicated by involved mucosal epithelium of the airway, a preoperative consultation with the anesthesiologist is recommended. Esophageal stricture may also be present.

The surgical goal is to improve function by placing the involved digits in a more favorable position. To achieve this goal, digits that are coalesced together from repeated scarring need to be separated, scars must be released, and wounds need to be covered (Fig. 50-4, A through C). Adequate separation of the digits may first require web space release of the adductor pollicis and incisions at the distal interphalangeal (DIP) and PIP flexion creases to allow extension of the digits. Wound coverage is often the most challenging aspect of surgical treatment. Coverage op-



Fig. 50-4 A-C, Intraoperative release of mitten deformity. The digits were separated, K-wires were placed to maintain separation, and Apligraf was used for wound coverage. D and E, Postoperative results after the separation of mitten deformity, K-wire fixation, and application of Apligraf.

tions have evolved, from local flaps and skin grafts, to leaving the wounds open, to allografts.⁴⁻¹⁰ Allograft types include allogenic composite cultured, acellular allograft dermal matrix, and living cell skin substitute (Apligraf)⁸⁻¹⁰ (Fig. 50-4, *D* and *E*). No one method of wound coverage has been found to be consistently reliable in the EB patient.

As with all hand procedures, the two opposing aspects of postoperative care—immobilization and motion—must be balanced. The postoperative dressing should maintain the position after separation. Skin care must be a priority at every stage of the postoperative period, from the dressing, to splint formation, to motion exercises. Motion exercises usually start 4 to 6 weeks after surgery, and night splints are worn for up to 3 months. If surgery results in improvements, it is difficult to predict how long they will be maintained. The severity of the initial deformities, the patient's adherence to postoperative splinting and therapy, and the patient's overall health and nutritional status are important factors in the outcome of surgery. Surgical intervention has been shown to improve grasp and gross pinch, but maintenance of these gains has been difficult.⁶ Any gains made at the time of surgery should be considered temporary, and recurrence of the deformity should be expected in the future. Lifelong wound care with chronic skin blisters, joint contractures, and pseudosyndactyly should be expected. Research efforts focused on gene transfer, protein-based, and stem cell-based therapies hold some promise but may not be clinically applicable for many years.¹¹

Contracture Disorders: Arthrogryposis

Arthrogryposis is actually a physical finding, not a diagnosis. It represents a large group of disorders, all of which are characterized by the presence of joint contractures at birth. Arthrogryposis is sometimes used as a noun to describe specific diseases. As an adjective, *arthrogrypotic* refers to rigid joint contractures. More than 65 distinct syndromes are coded under the term *arthrogryposis* in the Online Mendelian Inheritance in Man (OMIM) database,¹² illustrating the large variety of causes associated with this term. Although each condition has a different clinical course, prognosis, genetic basis, cause, and pathophysiology, confounding surgical decision-making for an individual patient, a simple way to think about these disorders is to consider them as contracture syndromes. They can then be grouped into a few general categories, each of which can be represented by a prototypical disease that can be managed with shared principles.^{13,14}

The contracture syndrome groups are as follows:

- Those involving all four extremities: This group includes arthrogryposis multiplex congenita and Larsen syndrome.
- Those involving, predominantly or exclusively, the hands and feet; these are the distal arthrogryposes. Facial involvement can occur with some of these syndromes, and Freeman-Sheldon “whistling face” syndrome is included.
- *Pterygia syndromes*, in which identifiable skin webs cross the flexion aspects of the knees, elbows, and other joints: Multiple pterygia and popliteal pterygia fit into this group.

Arthrogryposis Multiplex Congenita

Arthrogryposis multiplex congenita is the best known of the multiple congenital contracture syndromes.^{15,16} Although attempts have been made to change the name *arthrogryposis multiplex congenita* to *multiple congenital contractures* or *amyoplasia*, the popularity of the term *arthrogryposis* remains. The cause of arthrogryposis multiplex congenita is unknown. It was initially described in 1841 by Adolf Wilhelm Otto, who referred to his patient as “a human monster with inwardly curved extremities.”¹⁷ It is a sporadic condition, not an inherited one, and arthrogryposis may affect only one member of a pair of identical twins.^{18,19} It has been suggested that teratogens are



Fig. 50-5 PIP joint contractures in the hand of a patient with arthrogryposis. Note the lack of flexion creases at the PIP joints.

a cause of arthrogryposis, but this has not been proved.^{16,20-23} Some mothers of children with arthrogryposis have serum antibodies that inhibit fetal acetylcholine receptor function, raising the possibility that maternal antibodies play a role in the pathophysiology of these fetal antigens.²⁴

Histologic analysis of the muscles in this disorder shows fibrosis and fat between the muscle fibers, with myopathic and neuropathic features in the same muscle biopsy specimen. The peri-articular soft tissue structures are fibrotic. Analysis of the spinal cord shows a decreased number of anterior horn cells in the spinal cord.²⁵⁻²⁷ The pattern of motor neuron loss in specific spinal cord segments correlates with the peripheral deformities and affected muscles, suggesting a primary central nervous pathology in the etiologic factor.²⁸

The diagnosis can be made with clinical examination. The limbs are striking in appearance and position and lack normal skin creases (Fig. 50-5). There may be deep dimples over the joints. Muscle mass is reduced, although infants often have abundant subcutaneous tissue. Affected individuals hold the shoulders adducted and internally rotated, with the elbow more often extended than flexed, and the wrist flexed severely with ulnar deviation. The fingers are flexed and clutch the thumb. The hips are flexed, abducted, and externally rotated. The knees are typically in extension, although flexion is possible; feet often have a clubfoot disorder. Joint motion is restricted. There is a firm, inelastic block to movement. In two thirds of patients, all four limbs are affected equally, but in one third, lower limb deformities predominate; the upper extremities predominate only on rare occasions. There can also be dislocation of the hips.

Fifteen percent of children with arthrogryposis also have an inguinal hernia, although this is because of muscle weakness. Feeding difficulties can be encountered in infancy; these lead to respiratory infections and failure to thrive.²⁹ The face is not particularly dysmorphic.

Radiographs show relatively normal joints, which develop adaptive changes. Electromyograms (EMGs) and muscle biopsies have questionable diagnostic value. A diagnosis of arthrogryposis can be suspected when an absence of fetal movement is detected through prenatal ultrasonography, especially if this occurs in combination with polyhydramnios.²⁰

The natural history of arthrogryposis is not well studied.^{30,31} Some contractures seem to worsen with age, and the joint becomes stiffer. No new joints become involved. At least 25% of patients do not ambulate, whereas many others are limited to walking in the house.³² Patients with arthrogryposis who are very weak as infants remain weak throughout life, and those who appear stronger as infants stay strong. Adult function is more related to education and coping skills than to the severity of the joint contractures.

Treatment

Although there are potential surgical options for each joint, an overview of the total patient is essential when planning surgery. In the upper extremity, the overall goal is motion for self-care.^{13,33-35} Intraarticular changes occur between 4 and 6 years of age; outcomes of joint surgeries are often more successful if they take place before these changes. Realignment osteotomies, however, are usually performed closer to the completion of growth. Early motion and avoidance of prolonged casting may increase joint motion, thereby improving function. Long-term bracing or other assistive devices are often required.¹⁶

Because of joint contractures, the birthing process may be difficult for the infant, resulting in neonatal fractures.³⁶ Physical therapy should not be initiated in the newborn until such fractures are ruled out.³⁷ No studies clearly demonstrate that early mobilization improves patient outcome, but such a program is commonly prescribed and is thought to improve passive range of motion.¹⁶ Fractures may accompany an overly vigorous range-of-motion program.

Most patients do not require surgical procedures of the upper extremity. When evaluating a patient, one must consider an individual joint in the context of the entire upper extremity.^{29,30} Analysis must include an examination of each hand alone and how the two hands work together. Surgery should be undertaken to improve the use of the upper extremities in self-help skills, such as feeding or toileting, and mobility skills, such as pushing out of a chair or using crutches.

The shoulder rarely requires surgery. For the elbow, it is ideal to achieve flexion to 90 degrees from the fixed extended position. However, when both elbows are involved, surgery to increase flexion should only be done on one side at a time. Achieving active elbow flexion is unlikely because of contractures of the joint capsules and muscle weakness. Passive elbow flexion to a right angle is a prerequisite for considering a tendon transfer for active elbow flexion. Restoration of elbow motion by capsulotomy and triceps lengthening has had only fair success, diminishing the likelihood for success when an arthrogryptic muscle is used to motor the joint.³⁸ The triceps brachii and pectoralis are the most commonly used muscles for transfers. Surgery works best in children older than 4 years of age or in those who have at least grade 4 strength of the transferred muscle.³⁹⁻⁴³ Distal humeral osteotomy can be undertaken to place the elbow into flexion and correct some of the shoulder's internal rotation deformity. Osteotomy is usually performed when the patient is about 10 years of age.^{40,44} This surgery should be undertaken in a way that improves hand-to-mouth function. Excessive external rotation of the distal humerus should be avoided.

The hand and wrist are usually flexed and ulnarly deviated.^{45,46} All tissues of the wrist (joint capsule, tendons, fascia, subcutaneous tissue, and skin) are contracted. The ulnar deviation occurs because of unopposed action of the extensor carpi ulnaris, which is usually spared. Numerous techniques to place the wrist in a more neutral position have been described, including proximal row carpectomy, dorsal radial wedge osteotomy, arthrodesis, and resection of a biplanar wedge of midcarpus, with tendon transfer for wrist extension.⁴⁷ Results are unpredictable, and surgery at the wrist should not be considered without first evaluating the extremity as a whole. For example, wrist fusion would remove all wrist motion in a limb that may not have the compensatory abilities of forearm rotation and shoulder abduction. Thus the patient may be left with a less functional limb.

Ulnar-side digits tend to be more involved. PIP flexion deformities rarely respond to physical therapy or surgery. The thumb is flexed and adducted into the palm, and it responds better to surgery than do the other digits. Adductor release and web space deepening with Z-plasty can be performed. Children with arthrogryposis have normal intelligence and usually have a natural ability to learn substitution techniques.

Neuromuscular Disorders: Cerebral Palsy

The cause of cerebral palsy (CP) is unknown; initially, it was thought to be caused by prenatal or early postnatal insults to the CNS. However, this notion has been challenged by data, suggesting that the CNS insults occur much earlier in fetal development. Both the upper and lower extremities can be involved. The typical posture of the upper extremity is characterized by⁴⁸:

- Internal rotation of the shoulder
- Elbow flexion
- Forearm pronation
- Wrist flexion and ulnar deviation
- Thumb adduction or thumb-in-palm deformity
- Finger swan neck or clenched fist deformity (Fig. 50-6)

The prevalence of CP is estimated at 2.5 per 1000 live births, with low birthweight, intrauterine infections, and multiple gestation as the most important risk factors.⁴⁹

Because the cause of CP is unclear and the term may be used to describe conditions caused by several etiologic factors, it can be difficult to diagnose CP in young children. Thus it may be better to think of affected children as having a muscle spasticity disorder. In addition, before 1 or 2 years of age, upper extremity dysfunction may not be apparent. As a child reaches developmental milestones—the development of a pinch between the thumb and index finger, for example—the deficits become easier to diagnose. Because handedness usually does not develop until 18 months, a demonstrated preference for the right or left side before 12 months of age suggests a neurologic insult affecting the nonpreferred side.⁴⁸ Abnormal muscle tone can also suggest a diagnosis of CP, with muscle flaccidity and generalized hypotonia at birth progressing to hypertonia and contractures as the child becomes older.⁴⁸ Persistence of the primitive Moro reflex beyond 6 months, at which point it is normally lost, is another early sign suggestive of CP.⁵⁰

Approximately two thirds of children with CP are mentally impaired, with more than half of those patients moderately or severely mentally disabled.⁵¹ Although the patient's ability to participate in postoperative therapy is important for a favorable outcome, intellectual capacity is only one of many factors that are considered in the management of a CP patient's spasticity.

Four types of CP have been described by Hagberg et al.⁵² These are based on the type of movement disorder: (1) spastic, (2) athetoid, (3) ataxic, and (4) mixed. *Spastic CP* is the most common type, representing more than 80% of cases. Motor spasticity causes muscle imbalance across



Fig. 50-6 Typical posture of a patient with CP. Note the shoulder's internal rotation, elbow flexion, forearm pronation, and wrist flexion.

the joints of the upper and lower extremities, leading to stiffness and eventual contracture. *Athetoid CP* is reported in 10% to 25% of cases and is characterized by slow, withering movements and a lack of voluntary control. *Ataxic CP* is reported in 5% to 10% of cases and is also characterized by a lack of voluntary control; imbalance and tremors are characteristics of the condition. *Mixed CP* is composed of elements of more than one type. The incidence of patients with ataxic and athetoid CP has declined drastically in recent decades because of the recognition that it is caused by maternal-fetal blood cell-antibody mismatch.

Although a specific cause for CP has not been identified, multiple risk factors have been described. These include prematurity, teratogens, genetic syndromes, chromosomal abnormalities, exposure to toxins, intrauterine infections, intracerebral hemorrhage or stroke, complications during labor and delivery, and preeclampsia.⁵³ Early descriptions of CP often cited anoxic brain injury as the cause, but recent reviews of the literature have found that anoxia alone accounts for fewer than 10% of CP cases, and in most cases, a specific cause cannot be determined.⁴⁸

Treatment Goals

Both nonsurgical and surgical treatments are based on maximizing function of the extremity and allowing independence in daily living. In more severe cases, treatment may address hygiene problems from contracted joints. For each CP patient, the treatment plan must be individualized to fit that patient's needs. Perhaps the most difficult decision that a hand surgeon must make is selecting the appropriate patient for surgical intervention. Multiple issues must be considered, such as muscle weakness, spasticity, level of voluntary control, degree of joint stiffness or contracture, sensation, and patient intelligence. Because of this complexity, the hand surgeon must be viewed as a consultant in the overall team management of a CP patient. Input should be sought from all who care for a CP patient: the patient's parents, pediatrician or family doctor, rehabilitation specialist, physical therapist, occupational therapist, and surgeon. A treatment plan with realistic expectations can then be formulated.

Early management of a CP patient includes nonoperative treatments, such as stretching and splinting, which may preserve joint mobility and prevent contractures; however, there is no evidence that this improves patient outcome. Surgery is usually not considered until the patient is older, when independence and level of function can be more adequately assessed. If there is no further improvement or the patient's function worsens despite nonoperative measures, and if the patient can participate in postoperative therapy, surgery is considered.

Patient Selection

The decision to proceed with surgery requires assessment of intelligence, motor control, joint mobility, sensation, support for postoperative care, and level of function. Although many patients with CP are mentally disabled, this should not automatically eliminate a patient from consideration as a candidate for surgery. Optimizing hand position to improve function is an achievable goal, even in a patient with decreased intelligence. However, if mental status is such that participation in postoperative therapy is not possible or the patient demonstrates neglect of the extremity, surgery will not provide much functional benefit. Similarly, if the patient demonstrates a complete lack of sensation, it is unlikely that any surgery would allow the patient to incorporate and use that hand.⁵⁴ In patients in whom addressing long-standing contractures is related more to facilitating hygiene rather than to improving function, intelligence and sensation are less of an issue.

Motor control and joint status must also be assessed. In a CP patient, muscles may be spastic, flaccid, or dyskinetic, such as in the patient who is athetoid. Joint deformity is a result of the muscle imbalance caused by spasticity. Surgery to improve position and function is based on the rebalancing of forces across the joint. This can be accomplished by either reducing the force of the spastic muscles or increasing the force of the antagonist muscles with the use of appropriately selected tendon transfers. Athetosis is considered a contraindication to surgery, because the lack

of voluntary motor control makes muscle rebalancing difficult; this may lead to an unpredictable and unsatisfactory result.

EMG testing may be helpful to identify spastic or flaccid muscles for tendon transfer surgery. Videotaped assessment of range of motion and function of the upper extremity has been found to be a reliable assessment measure.⁵⁵ Therapist-assisted videotaped evaluations can be useful in helping the surgeon develop the optimal treatment plan. Information obtained from repeated viewing of the evaluations adds to the clinical picture provided by the history and physical examination, and it may allow the surgeon to develop a more thoughtful and effective treatment plan.⁵⁵ The videotaped evaluation should include assessment of range of motion of the upper extremity, presence of any joint contractures, and level of function. To assess function, the Jebsen hand function test can be administered, and manual dexterity tasks can be demonstrated.⁵⁵ Both EMG and videotaped assessments should be used as adjuncts to a thorough history and physical examination in patient selection for surgery.

Nonsurgical Treatment

Splinting is often prescribed to prevent the development of joint contractures. The use of splints is somewhat controversial, because decreased spasticity and prevention of contractures have not been shown with splinting.⁵⁶ In addition, splinting may actually enhance the deformity by placing a hyperexcitable muscle under tension and increasing muscle tone.⁵⁴ Nevertheless, splints are used to improve hand position and may help to perform activities of daily living. If used at night when the child is completely relaxed, splints may help to prevent contractures.⁴⁸

The use of botulinum toxin (Botox) to decrease motor spasticity in a CP patient is becoming more widely reported.⁵⁷ It is more commonly used for lower extremity spasticity. The goal of botulinum toxin therapy is to improve function by decreasing muscle tone. However, the long-term effectiveness of botulinum toxin in a CP patient has not been shown, and indications for use have not been clearly established. In the treatment of ankle equinus contractures, the addition of botulinum toxin to a serial casting regimen led to an earlier recurrence of spasticity, contracture, and equinus during gait when compared with the casting-only group of patients.⁵⁷

In our experience, Botox treatment works best in younger children without fixed contractures or in those who have limitations in function caused by dynamic spasticity with active motion. This is the same group of patients in whom splinting may produce the most functional improvement.

Surgical Treatment

Surgical treatment is based on addressing the patient's motor spasticity and subsequent muscle imbalance, which are responsible for producing the characteristic joint deformities. Surgical options to correct the imbalance by weakening the spastic muscle include muscle denervation, muscle or tendon lengthening, and muscle release. Surgical options include tendon transfers and tenodesis to correct the imbalance by augmenting the power of a muscle opposing the spastic muscle. If the surgical goal is to place the hand in a more functional position to facilitate skin care, joint fusion is an option.

Selecting which patients will benefit from surgery is more challenging than deciding on the type of surgery. Surgery should only be undertaken to correct a specific functional defect. In our experience, most CP patients do not require surgical treatment.

The typical posture of the upper extremity is characterized by:

- Shoulder internal rotation
- Elbow flexion
- Forearm pronation
- Wrist flexion and ulnar deviation
- Thumb adduction or thumb-in-palm deformity
- Finger swan neck or clenched fist deformity

Patients will present with variable severity at multiple joints. No one technique can be universally applied to all CP patients, and treatment plans must be individualized based on careful functional assessment and input from all members of the multidisciplinary team. If a patient is considered a good surgical candidate, corrective surgery is planned to address multiple joints in as few visits to the operating room as possible to minimize the anesthetic risk and postoperative morbidity.

Elbow flexion contracture is a common finding in CP patients because of the spasticity of the biceps, brachialis, and flexor-pronator muscles. Patients may have difficulty with feeding and self-care because of the position of the hand when the elbow is fixed in flexion. Transfers to a chair, keyboarding, and using assistive devices at tabletop level may be more difficult with an elbow flexion contracture. In more severely debilitated patients, a flexion contracture may lead to hygiene problems and chronic wounds in the antecubital fossa. Fractional lengthening or step cuts allow lengthening of the biceps and brachialis tendons.^{54,58,59} Release is achieved through a zigzag or lazy-S incision in the antecubital fossa. The median nerve, brachial artery, and biceps and brachialis tendons are identified. Surgical release should be done cautiously, because it may weaken elbow flexion to the point where the extremity is in a more extended but less functional position for feeding and self-care activities. In contractures that have been present for a long time, joint capsule release may also need to be done to improve elbow position.

Pronation is the usual posture of the forearm because of the spasticity of the pronator muscles and weakness of supination. Excessive forearm pronation affects self-care, feeding, and use of a keyboard or communication board. Techniques to correct the pronation imbalance and place the forearm in a more neutral position include pronator fasciectomy, pronator lengthening, and pronator rerouting. Gschwind and Tonkin⁶⁰ described pronator quadratus fasciectomy for mild contracture. The pronator teres can also be lengthened by transecting it or doing a step cut at its insertion on the radius.⁶¹ The tendon ends are sutured together with the forearm in maximal supination. If supination is weak or absent, the pronator teres can be released with a strip of periosteum off its insertion, rerouted through the interosseous membrane and around the radius, and then reinserted onto the radius to serve as a supination force against the pronator quadratus.^{60,62}

The wrist is typically flexed in a CP patient because of the spasticity of the wrist flexors, which are often combined with weak extensors. A flexed wrist interferes with activities of daily living, such as eating and drinking, self-care, dressing, and typing on a keyboard. Grip strength is decreased if the wrist is in a flexed position. Passive range of motion to the neutral position must be achieved before rebalancing surgery is considered. Wrist flexion contracture can be improved by lengthening the flexors, releasing the wrist capsule, and performing tendon transfer to augment wrist extension. Tendons that serve as good donors for transfer in non-CP patients may not be good sources in a CP patient because of a global neuromuscular disorder. Wrist extension must be evaluated independent from and in conjunction with finger extension.

Procedures that can be used to correct wrist flexion contracture include flexor fasciectomy, flexor tenotomy, flexor tendon lengthening, and superficialis-to-profundus transfer. In mild cases of contracture, the fascia can be excised through a longitudinal incision on the palmar aspect of the forearm. Tenotomy of the flexor carpi ulnaris (FCU) can be done just proximal to its insertion on the pisiform to allow more wrist extension.⁶³ If tenotomy of the FCU does not produce significant improvement, tenotomy of the flexor carpi radialis may be considered.⁴⁸ However, Tonkin and Gschwind⁶³ considered a lack of voluntary control of the radial wrist extensor a contraindication to tenotomy of the FCU and did not recommend tenotomies of both wrist flexors. Flexor tendon lengthening by step cuts or fractional lengthening is considered when full active and passive extension of the fingers is demonstrated only when the wrist is flexed 20 degrees or more. Improvement in grip may be seen in flexor tendon lengthening, because it may place the wrist in a more extended position. In more severe cases of flexor tendon contracture—those in which improvements in hygiene and appearance are more achievable than improvement in functional grasp—*superficialis-to-profundus* (STP) transfer can be done.⁶⁴ In this procedure, the

superficialis tendons are divided proximal to the wrist, and the profundus tendons are cut near the musculotendinous junction. The proximal cut ends of the superficialis tendons are then woven into the distal cut ends of the profundus tendon. With this transfer, a tenodesis effect is produced, with the fingers first extended during wrist flexion and then flexed with wrist extension.

Weak wrist extension can be improved with tendon transfer, usually of the FCU. The FCU is a contributing factor in a flexion contracture. Release of the FCU and transfer to the extensor carpi radialis brevis (ECRB) tendon accomplishes two goals in rebalancing the forces across the wrist by weakening flexion and strengthening extension. The FCU is the preferred donor for extension of the wrist in the CP patient, because it is typically available and usually provides a reliable result.⁶⁵ The technique of FCU-to-ECRB transfer involves releasing the FCU tendon from the pisiform and then rerouting it around the ulna to the ECRB tendon on the radial side. The FCU muscle is dissected free from surrounding fascia to allow adequate mobilization for the tendon weave. This mobilization allows proper alignment of the musculotendinous unit and maximizes the force of the transfer.

Before correcting the imbalance at the wrist by augmenting extension, the effect of the transfer on finger extension must be considered. The pronator teres and the brachioradialis can be transferred to the ECRB for active wrist extension. In a CP patient, these transfers are less reliable and may produce a permanently extended wrist, thereby decreasing grasp power and impeding release of the hand. Range of motion of the digits must be assessed with the wrist. If full passive extension of the digits is possible and weak release of the digits is the main problem, then FCU-to-extensor digitorum communis (EDC) transfer should be considered to augment active digital extension. If passive extension is limited, however, then fractional lengthening of finger flexors should be considered. In the severely contracted wrist, release of all the wrist flexors (FCU, flexor carpi radialis, and palmaris longus) may not be enough to place the wrist in a neutral position, and wrist fusion will be required.

The thumb often demonstrates an adduction contracture, with the thumb metacarpal adducted against the palm or second metacarpal. The imbalance of forces can also produce *metacarpophalangeal* (MP) or IP joint instability. Spasticity of both the adductor pollicis and the first dorsal interosseous muscles, as well as the weakness of the thumb abduction, also contribute to the characteristic deformity (Fig. 50-7). Surgical treatment of the thumb addresses the adduction contracture and joint instability. Release of the contracture is achieved by a dorsoulnar incision to expose the first dorsal interosseous muscle and adductor pollicis. The fascia of the first dorsal interosseous muscle is excised, and in some patients, the muscle may need to be released



Fig. 50-7 Thumb adduction contracture in a patient with CP. Spasticity of the adductor pollicis and first dorsal interosseous muscles, weakness of thumb abduction, MP joint instability, and tightness of the first web space contribute to this deformity.

from its insertion on the first metacarpal. The adductor pollicis is lengthened by detaching it in step-cut fashion and then suturing it to itself. More palmar dissection allows release of the adductor pollicis origin from the third metacarpal.⁶⁶ The first web space skin contracture can be lengthened by Z-plasty. Shortening of the abductor pollicis brevis (APB) tendon or transfer of an active motor, such as the brachioradialis, can be done to augment weak thumb abduction. To improve thumb extension, the extensor pollicis longus (EPL) can be transected at the MP level, passed through a separate dorsal wrist incision, rerouted through the first dorsal compartment, and then sutured back into place at the MP joint. Less common than adduction contracture is the thumb-in-palm deformity, which is produced by a spastic flexor pollicis longus (FPL) and weak thumb extension: EPL, extensor pollicis brevis (EPB), and abductor pollicis longus (APL).⁶⁷ Options to correct this imbalance include FPL lengthening and flexor digitorum superficialis (FPL; fourth digit) transfer to the EPL.⁶⁸

The characteristic finding of the fingers is the swan neck deformity, typically caused by intrinsic muscle spasticity, although the long extensors can be shortened in a long-standing wrist flexion contracture. Because the swan neck deformity can be improved with corrective surgery for the wrist flexion contracture, surgery to correct the swan neck deformity is usually delayed until the effect of the wrist rebalancing surgery is known. Surgical options to address the swan neck deformity include superficialis tenodesis, intrinsic muscle denervation, tendon transfers, lateral band tenodesis, PIP palmar plate arthroplasty, and PIP joint fusion.⁶⁹⁻⁷¹

Postoperative management for tendon transfers involves immobilization for 4 to 6 weeks, followed by exercises to increase range of motion. Night splints are worn for approximately 3 months after surgery. Splints may also be used beyond the 3-month period to provide support or prevent new deformity.

Inflammatory Disorders: Juvenile Rheumatoid Arthritis

Juvenile rheumatoid arthritis (JRA) is the most common connective tissue disease occurring during childhood, affecting approximately 60,000 to 250,000 individuals.⁷² JRA is characterized by chronic synovial inflammation and hyperplasia, which lead to edematous and hyperemic tissue grossly. On the microscopic level, there is infiltration by lymphocytes and plasma cells. Clinically, this continued inflammation may result in progressive periarticular erosions and joint destruction. Compared with the adult form of rheumatoid arthritis (RA), JRA has a much milder course, with approximately 50% to 70% of cases going into remission.⁷³ There are three types of JRA: (1) systemic onset (Still's disease), (2) polyarticular, and (3) pauciarticular.

Clinical features of *Still's disease* include:

- Intermittent high fevers
- Erythematous macular rash
- Generalized lymphadenopathy
- Hepatosplenomegaly
- Leukocytosis
- Anemia
- Uveitis
- Arthralgia
- Myalgia
- Transient arthritis

Rheumatoid factor (RF) and antinuclear antibodies (ANA) are usually not found. Of those patients with Still's disease, 25% have severe chronic arthritis.⁷² Approximately 40% of patients have the polyarticular form of JRA. *Polyarticular JRA* most resembles adult RA and is also the

most likely to progress to adult RA. It is characterized by multiple joint involvement and occurs more frequently in females than in males. Most patients with polyarticular JRA are not RF positive, but those who are RF positive are more likely to develop severe chronic arthritis as adults.⁷²

The remaining 40% of cases are *pauciarticular JRA*, which is characterized by the involvement of four or fewer joints within the first 6 months of the onset of symptoms. Two distinct groups are affected. The first group is characterized by female patients younger than 6 years of age who are usually ANA positive and RF negative. Joint involvement of the lower extremity and iridocyclitis are more common in this group. Other joints may become involved later in the course of the disease. The second group is older and predominantly male, with an association with the HLA-B27 haplotype. These patients are ANA and RF negative. The lower extremity joints are also involved in this group.⁷³

To establish a diagnosis of JRA, other inflammatory conditions, such as septic arthritis, osteomyelitis, inflammatory bowel disease, and malignancy, should be ruled out. Unlike adults, RF may not be found in JRA patients or may be seen only in older pediatric patients. The absence of RF in a child with clinical signs of JRA does not exclude the diagnosis.

The number of affected joints and the severity are variable. In the hand, the characteristic findings are loss of flexion and the presence of radial deviation at the MP joints and a loss of IP joint flexion. In an adult, the characteristic finding is ulnar deviation and loss of extension at the MP joint.⁷³ At the wrist, the typical presentation is ulnar deviation and loss of wrist extension. There may also be ulnar shortening up to 9 mm, but this does not correlate with ulnar deviation or MP radial deviation.⁷² Proliferative synovitis may lead to joint edema and stiffness and subsequent loss of flexion. Local synovitis can also cause epiphyseal overgrowth or undergrowth around affected joints and may result in premature physal closure. The resultant bony deformity is affected by the duration of the disease. If the onset of disease occurs before skeletal maturity, growth of the hand may be diminished.

Radiographic findings can demonstrate early signs of disease with periarticular soft tissue swelling, juxtaarticular osteoporosis, and periostitis. Radiographic findings of late disease include destruction of joint, cartilage, and bone; bony fusions; and growth anomalies, such as premature epiphyseal closure.⁷² Bony change is most commonly seen at the knee and wrist. A shortened ulna and abnormal growth of the fourth and fifth metacarpals from premature physal closure may be seen. There may also be narrowing of the intercarpal, radiocarpal, and carpometacarpal joints, diminished size of the small bones of the hand, or premature ossification of the wrist bones.

After the diagnosis of JRA is made and the subtype is identified, treatment goals are directed at minimizing symptoms, preventing deformity, and preserving function. As with other chronic conditions, care is supportive rather than curative. A multidisciplinary approach involving the pediatrician, rheumatologist, physical therapist, occupational therapist, orthopedist, and hand surgeon is advocated.

Nonsurgical Treatment

Nonsurgical treatment is directed at relieving symptoms, maintaining good joint position, preserving function, facilitating activities of daily living, and preventing deformities. First-line therapy includes antiinflammatory medication, physical therapy, and splinting. As with adults, medical therapy can be the most effective way to control disease. Pharmacologic options include aspirin, NSAIDs, gold, D-penicillamine, methotrexate, sulfasalazine, and steroidal agents. NSAIDs are usually not prescribed on a regular basis until a patient is 14 years of age.⁷² Resting splints may provide comfort and help to place the hand in a more functional position. With improvements in pharmacologic management, the need for surgery has substantially declined. Novel therapies on the horizon may decrease the need for surgery even further in the future.

Surgical Treatment

In general, surgery of the upper extremity is rarely indicated in the JRA patient and is considered only after the use of nonoperative measures.⁷⁴ Reconstructive procedures commonly used in the adult RA population are not indicated in younger patients because of the risk of epiphyseal arrest resulting from injury to the growth plate. Surgery can be considered during the late teen years, when growth is completed and symptoms warrant surgical intervention. Patients who are RF positive are more likely to need surgery to alleviate symptoms.⁷⁴ Postoperative improvement in a JRA patient results from excision of proliferative synovitis and better joint motion. Affected joints are either stabilized or reconstructed.

The following issues should be considered before surgery: the pattern of joint involvement, the degree of skeletal immaturity, the timing of interventions, and the risk of surgery causing epiphyseal arrest. The goals of surgery are to alleviate pain, retard progression of disease, improve function, improve appearance, and maintain growth and motion.⁷² At the wrist and MP joints, synovectomy can be done to decrease the inflammatory activity at the joints and prevent its sequelae of joint destruction and fixed deformity. Hand and wrist involvement in the JRA patient has been reported in up to 59% of patients.^{73,75} Synovectomy is usually considered after 6 months of persistent synovitis and failure of medical management. Candidates for surgery usually present with swollen, painful, boggy joints, decreased motion, and radiographic evidence of joint destruction. As with surgery in the hand, there is a risk of inducing more swelling, stiffness, and decreased motion postoperatively. This risk must be weighed against the potential benefit of the surgery to relieve pain, which is reported in the population.⁷²

There are numerous well-described procedures to address the problems of the small joints of the hand: boutonnière deformity, swan neck deformity, palmar plate laxity, dorsal capsule tightness, and collateral ligament shortening. The most common procedures in the hand for the JRA patient are MP joint capsulectomy for MP joint contracture, sublimis resection for PIP joint contracture, and ulnar collateral ligament (UCL) tightening. Indications and techniques for these surgeries are the same as in an adult RA patient, although they are performed on a much less frequent basis. Joint replacement is not advocated in a JRA patient because of the limited life expectancy of the implant and the high demands of younger, more active patients.

Hematologic Disorders: Fanconi Anemia

Fanconi anemia (FA) is an autosomal recessive disorder of bone marrow dysfunction that is associated with multiple birth defects, including upper extremity anomalies. The preaxial structures—the thumb and radius—are the most commonly affected and represent both cosmetic and functional deformities. The most common anomaly seen in patients with FA is thumb hypoplasia and radial dysplasia, followed by thumb hypoplasia alone and radial polydactyly.⁷⁶ Other clinical features include short stature, café-au-lait spots, red cell macrocytosis, and renal, genitourinary, and cardiac anomalies. It is critical to distinguish FA from thrombocytopenia absent radius (TAR) syndrome. Hematologic dysfunction in FA worsens with time, so surgery should be considered early in life; with TAR syndrome, the hematologic deficits improve over time, and therefore surgery should be delayed. One can easily differentiate between the two conditions, because patients who have TAR always have a fully formed thumb.

A patient with a thumb anomaly may present to a hand surgeon without any workup for FA. If other clinical features of FA are present, the hand surgeon may initiate the evaluation of FA by referring the patient to a hematologist or pediatrician for additional testing. Early diagnosis allows proper management of the hematologic disorder, appropriate timing of any elective surgery before the advent of bone marrow failure, and genetic counseling for the family. The median age at which hematologic complications occur is 8 years, but there is a wide range.⁷⁷

The goal is to maximize hand function and promote independence within the context of a chronic hematologic disorder. If the underlying blood disorder is under control or goes into remission, surgery can be considered. One of the goals of the treatment team may be to complete any surgeries to improve function of the hand before the development of any hematologic problems.

Because there is a wide spectrum of presentation, there is no single best approach or technique to manage these patients. The treatment plan must be individualized, taking into consideration the patient's overall function, the potential benefit of the procedure, and input from physical and occupational therapists.

After the decision is made to proceed with surgery, hematologic issues must be addressed. Blood work is done to see whether any coagulopathies are present and to check for evidence of thrombocytopenia, leukopenia, and anemia. In the patient with FA, laboratory tests may help to decide whether to delay surgery or whether it is safe to proceed. There should be a preoperative assessment by both the physical therapist and occupational therapist. The workup may also include investigations for cardiac, renal, and genitourinary anomalies, if indicated.

Involvement of the thumb and radius is variable, ranging from hypoplasia to absence. Evaluation and treatment of thumb hypoplasia are discussed in Chapter 43. Radial deficiency can present as a short radius (type I), a hypoplastic radius (type II), a partially absent radius (type III), or a radius that is completely absent (type IV) (Fig. 50-8). Complete absence is the most common



Fig. 50-8 Clinical appearance of total absence of the radius in a 5-year-old child. The presence of a thumb determines whether this condition is more likely Fanconi anemia (in which case the thumb would be hypoplastic or absent) or TAR syndrome (the thumb would be present).

Fig. 50-9 Radiograph of a 10-month-old patient with total absence of the radius. The thumb is present.



type of radial deficiency; in the FA patient, complete absence of the radius may also occur with complete absence of the thumb.

The typical posture in partial or complete absence of the radius is severe radial deviation of the wrist, stiff fingers, lack of forearm rotation, and limited elbow flexion (Fig. 50-9). Stretching is started at an early age, and splints are used to maintain a straight alignment. If the wrist continues to deviate radially despite nonoperative measures, then surgery is considered to improve function. Centralizing the wrist involves placing the carpus on the ulna. Although this provides a correction, recurrence is common.

KEY POINTS

- A multidisciplinary approach is needed for treatment of these complex patients.
- The entire upper extremity—not just the hand—must be assessed in these children to ensure proper potential positioning for effective use.
- The fragile skin in epidermolysis bullosa makes it susceptible to blistering and scarring, even with minor trauma. Great care must be taken with splinting, rehabilitation, and the administration of general anesthesia.
- Unfortunately, EB often progresses to irreversible pseudosyndactyly, mitten and cocoon hand, joint contractures, and muscle atrophy.
- Surgical goals in EB hand reconstruction are directed at positioning digits in a more functional position.
- Arthrogryposis is a physical finding of joint contractures at birth; these contractures can be seen in several syndromes and conditions.
- The cause of cerebral palsy remains unclear.
- The surgical management of CP is focused on maximizing function, allowing increased independence and use, and facilitating hygiene. Each surgery must be individualized to the child's physical, psychosocial, and environmental needs.
- Release of the thumb adduction contracture can be very rewarding—it may increase hand function significantly.
- The treatment goals for patients with juvenile rheumatoid arthritis are focused on relieving symptoms, preventing ongoing deformity, preserving or increasing function, and improving appearance.

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51

Toe-to-Hand Transfers in Children

Patrick L. Reavey • Neil F. Jones



oe-to-hand transfer is one of the triumphs of modern surgery and has been applied successfully to restore hand function in both adults and children after traumatic loss or congenital absence of digits. In children with congenital differences, toe-to-hand transfer has revolutionized hand reconstruction, and the lives of these children and their parents have been transformed by this marvel of modern medicine. The technique has been refined over the past five decades and is currently used throughout the world, primarily in major academic institutions and hospitals. It is a procedure with many risks and requires a dedicated team of microsurgeons, pediatricians, intensive care nurses, pediatric nurses, pediatric occupational hand therapists, and psychologists.

Nicoladoni¹ successfully completed the first toe-to-hand transfer in a child in 1898 using a staged pedicle transfer by joining the hand and foot together for 4 weeks. Other surgeons repeated the technique, but because of potential complications such as sciatic nerve palsy, pressure sores, and deep venous thrombosis, the concept of a one-stage free microvascular digital transfer was investigated. In 1964 Buncke et al² experimentally transferred a toe to the hand of a monkey and opened up the possibility of using this technique in humans. In 1966 Yang Dong-Yue of Shanghai First Medical College performed the first microvascular transfer of a second toe to the hand in a human.³ Cobbett⁴ was the first to transfer a great toe based on the plantar arterial system to reconstruct a thumb, and Buncke et al⁵ transferred the great toe based on the first dorsal metatarsal and dorsalis pedis arterial system. The pioneering work of these surgeons provided the basis for the widespread expansion and evolution of this reconstructive technique. Today many different centers have reported successful transfer of the great toe and second toe, simultaneous double toe transfer, staged double toe transfer, and vascularized joint transfers.⁶⁻⁵²

Relative to adults, toe-to-hand transfer is performed less frequently in children. However, with refinement in microsurgical techniques, the indications in children are increasing, and the success rate is similar to adults. Toe transfers have been used to reconstruct children's hands with missing digits as a result of trauma, congenital constriction ring syndrome, transverse deficiencies, cleft hand, symbrachydactyly, and even radial club hand.^{3-11,12-41,45,50} Toe-to-hand transfer is usually reserved for children with severely aplastic hands with absence of multiple digits. Although toe transfer has been reported for the treatment of isolated thumb hypoplasia, the benchmark remains pollicization of the index finger.^{22,37,40-42,44,53} The great toe and all lesser toes, either individually or together, have been transferred to the hand in the pediatric population.^{19,23,39}

Microsurgical toe transfer requires meticulous technique and a keen eye for aesthetics. It combines the fields of plastic surgery, orthopedic surgery, and neurosurgery in a way that is unparalleled in any other surgical endeavor. It is an important technique in the armamentarium of hand surgeons, but it will undoubtedly undergo continued refinement to optimally replicate the appearance and complex function of a human thumb or finger.

CONGENITAL HAND DIFFERENCES

The incidence of a child being born with absent or hypoplastic digits is approximately 1 in 6800 births.¹⁹ The diversity of congenital hand differences may at first seem overwhelming; however, they can usually be divided into two broad categories: transverse and longitudinal deficiencies and then further subdivided based on the presence or absence of particular digits. Transverse deficiencies may present as a true transverse arrest, congenital constriction ring syndrome, or symbrachydactyly (atypical cleft hand).^{1,7,18} Longitudinal deficiencies may present as thumb hypoplasia, radial club hand, cleft hand, or more rarely, ulnar club hand. Numerous anatomic and embryologic classification systems have been described for each of these processes. However, Jones and Kaplan⁵⁴ have described a unifying classification system based on the morphologic and radiographic appearance of congenital differences⁵⁴ (Fig. 51-1). This system provides a useful algorithm for approaching congenital hand reconstruction with either conventional or microsurgical techniques. Only the phenotypes R1 (distal to the carpometacarpal joint), R2, R3, R4, R5, U4R1, and occasionally C3R1U1 (at the proximal phalangeal level) are candidates for microsurgical reconstruction with toe-to-hand transfers.

In general, the anatomic structures in the hand are more developed in children with congenital constriction ring syndrome (see Fig. 51-5, *A* and *B*) than in children with hypoplasia, true transverse arrest, and symbrachydactyly^{3,19} (see Figs. 51-9, *A* and *B*, 51-10, *A* and *B*, and 51-11, *A* and *B*). Children with longitudinal deficiencies (see Fig. 51-8, *A* through *C*) may lack adequate proximal tendons and nerves necessary to power the transfer and may be more appropriate candidates for reconstruction by transfer of adjacent digits.^{7,49} In the monodactylous form of symbrachydactyly, a relatively consistent anatomy with the presence of carpal bones but loss of the central metacarpal rays is expected. The border metacarpals are usually present, but the thumb is often hypoplastic, unstable, and short or totally absent. Remnants of the fingers may be represented as small nubbins with or without nails.¹⁹

Symbrachydactyly is sometimes difficult to distinguish from simple transverse absence or transverse arrest. The small nubbins with nail remnants seen in symbrachydactyly are usually replaced in transverse arrest by small protuberances, dimples, or large fleshy bags of skin without skeletal contents in transverse arrest.⁷ The digital nerves and arteries may be very hypoplastic in an adactylous or monodactylous hand, requiring more proximal dissection to larger digital nerves for nerve repair and to the radial or ulnar artery for vessel anastomoses.

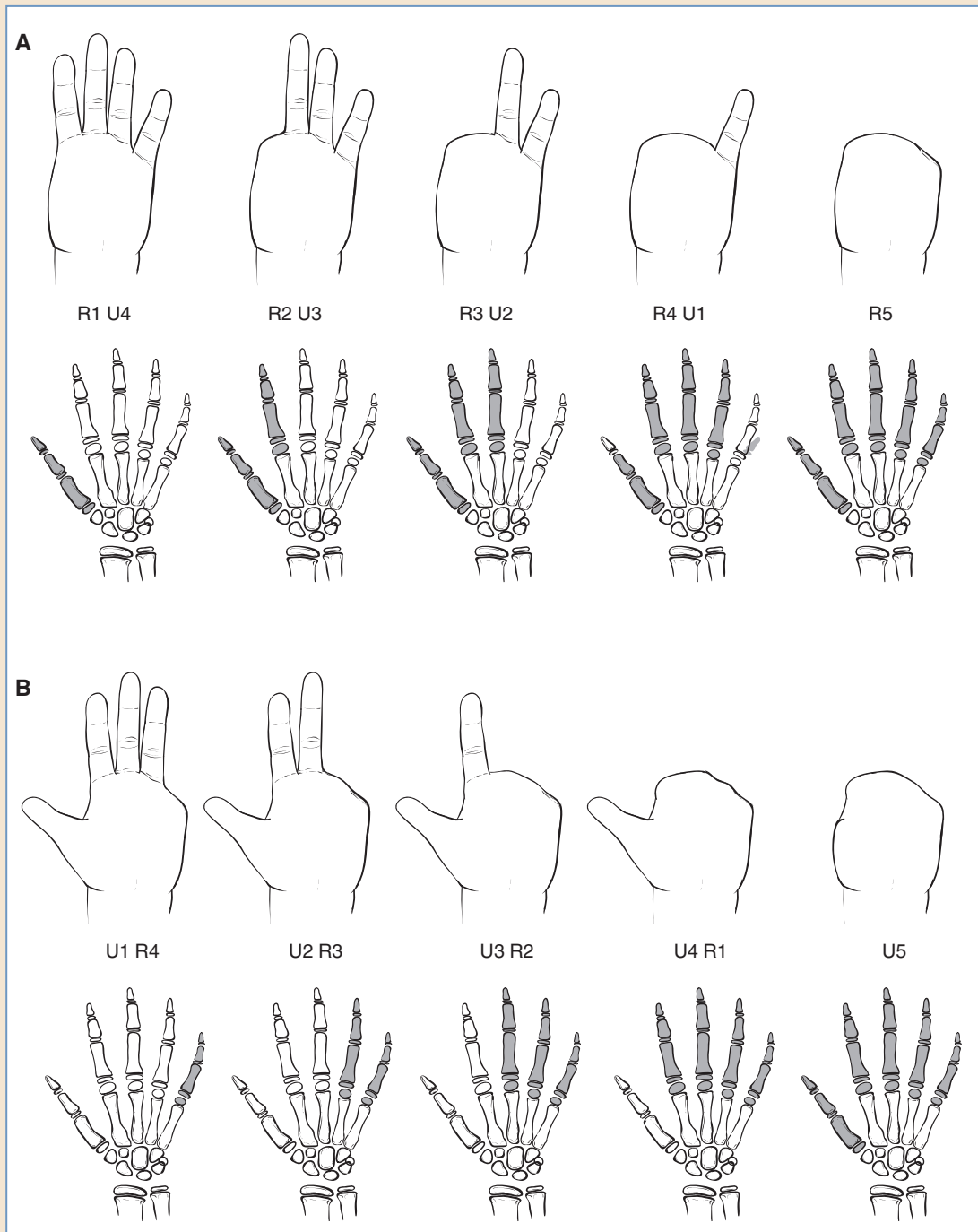


Fig. 51-1 Classification system for congenital absent digits: morphologic and radiographic spectrum. **A**, Radial deficiencies. **B**, Ulnar deficiencies.

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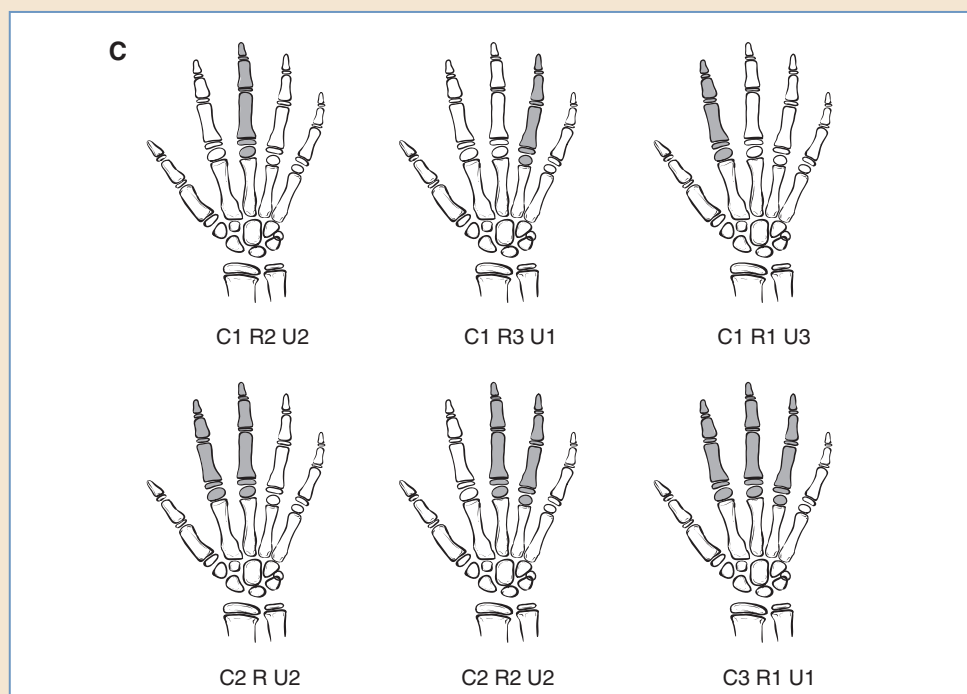


Fig. 51-1, cont'd C, The radiographic spectrum of central deficiencies. Only the phenotypes R1 (distal to the carpometacarpal joint), R2, R3, R4, R5, U4R1, and occasionally C3R1U1 (at the proximal phalangeal level) are candidates for microsurgical reconstruction with toe-to-hand transfers.

PREOPERATIVE ASSESSMENT

The initial assessment of a child with a congenital hand difference begins with careful history-taking, listening to the parents, and allowing them to express their understanding of their child's condition and their hopes and expectations for treatment. It has been shown that the parental response to a child born with a congenital anomaly resembles the bereavement response.^{44,55} Parents must cope with their own sense of loss, as well as the obligation to direct the treatment of their child. Some authors recommend that the parents and child meet with a clinical psychologist before considering a toe-to-hand transfer, and it has been shown that psychological counseling may play an integral role in decision-making in pediatric hand surgery.^{21,25} Additionally, several parents of children who have undergone a toe-to-hand transfer are willing to speak with families considering the operation. This provides an opportunity for the parents to ask questions about the entire reconstructive process and assuage some of their concerns in a way a medical professional might not.

Surgeons should obtain information regarding any significant past medical history that may indicate whether a child has a syndrome and whether anyone in the family has a similar hand condition. They should attempt to assess the functionality of the hand in a detailed physical examination. Observing very young children at play may be sufficient. Children should be examined with their shirt off to check for spinal and shoulder girdle abnormalities (Poland syndrome). The presence or absence of the thumb and its functionality are recorded. The thumb carpometacarpal joint is assessed, because it is critical to the success of a toe-to-thumb transfer.⁹ The presence or

absence of the radiocarpal joint and the number and size of the metacarpal bones is determined. Dimpling of the skin over the ends of the metacarpals with hand motion may indicate the presence of tendons that are tethered beneath the skin.

The feet are examined at the initial assessment. Congenital differences of the lower extremities may coexist with upper extremity differences.¹³ If possible, the foot may be brought close enough to the hand so that the parent and child can visualize what a transferred toe may look like. Parents usually greatly appreciate the chance to look at preoperative and postoperative molds and/or photographs and videos of toe transfers.

Preoperative imaging studies include plain radiographs of both hands and wrists and plain radiographs of both feet. The presence of metacarpals and of a thumb carpometacarpal joint is assessed; if this joint is absent, the surgeon may need to perform a pollicization of an adjacent metacarpal to serve as the base for the transferred toe. In cases in which the carpal and/or metacarpal bones have not yet ossified, an MRI may be useful.¹⁸

The need for preoperative angiograms of either the upper or lower extremity has been debated. Greenberg and May⁵⁶ found that preoperative lateral angiograms of the foot were helpful in identifying the location and size of the metatarsal arteries to the great toe and used angiography in all their cases. Multiple studies have shown significant variability in the dominant vasculature to both the great and second toes^{57,58} and variability between left and right sides.⁵⁹ However, most authors today agree that a preoperative arteriogram is not necessary, because suitable vessels for anastomosis can be found in all cases^{7,12,18} if the surgeon has a thorough understanding of the variations in arterial anatomy.

Congenital hand differences can be associated with abnormalities in other body systems. Radial longitudinal deficiency may be associated with cardiopulmonary, hematopoietic (Fanconi anemia and thrombocytopenia-absent radius [TAR] syndrome), and gastrointestinal and genitourinary abnormalities.⁶⁰ On the other hand, ulnar longitudinal deficiency presents more often with other musculoskeletal defects.⁶⁰ Cleft hand is associated with several syndromes, as well as cardiac, gastrointestinal, ocular, auditory, and skeletal abnormalities.⁷ The pediatrician usually performs an ultrasonographic examination of the heart and/or kidneys before the child's consultation with the hand surgeon. The pediatrician should always perform a preoperative evaluation before toe-to-hand transfer. For an operation that lasts 6 to 12 hours, children should be examined 1 to 2 days before surgery to rule out viral illnesses, which could compromise respiratory function.

TREATMENT

Nonsurgical

Prosthetic hands, fingers, and fingertips have been perfected to the point that they are virtually unnoticeable to the untrained observer. In an older child with a unilateral defect and a normal or near-normal thumb, a prosthesis may provide the best appearance and modest function.³

Surgical Indications

The most common indication for toe transfer in adults is traumatic amputation.⁶¹ In children this can be an indication for a toe transfer (Figs. 51-2 through 51-4), but more frequent indications are a congenitally absent or hypoplastic thumb, the absence of more than two fingers, or a combination of thumb and finger deficiency.³ Box 51-1 lists specific indications for a pediatric toe-to-hand transfer for both thumb and finger reconstruction.⁴⁷

The decision to perform a toe-to-hand transfer in a child involves a complex process between the surgeon, the parents, and the child. Whether a child is a candidate for a toe transfer and which specific toes should be transferred depends on the functional deficit, the appearance, bilateralism, the age of the patient, and the recipient anatomy.



Fig. 51-2 A, This 4-year-old boy had amputations of all five digits of his left hand in a lawn mower accident. B and C, After debridement and closure of the amputation stumps, he had only a remnant of the thumb metacarpal but still had no functioning thenar muscles. D-F, After a right second toe-to-thumb transfer, he has regained excellent opposition of the thumb and pinch and grasp. G, The donor site in his right foot is relatively inconspicuous.

Box 51-1 Indications for Pediatric Toe-to-Hand Transfers**Absent Thumb**

- Isolated absence of the thumb, distal to the metacarpal base, with a preserved carpometacarpal joint and thenar muscles and with four normal or relatively normal fingers
- Absence of the thumb, index, middle (and ring) fingers, with one or two fingers remaining on the ulnar side of the hand
- Unilateral and, extremely rarely, bilateral absence of all five digits

Absent Fingers

- Absence of all four fingers proximal to the base of the middle phalanges, more often proximal to the metacarpophalangeal joints, with a normal thumb
- Complete absence of all five digits. This can be reconstructed with two second toe transfers into the thumb and ring or small finger positions.



Fig. 51-3 A, This 7-year-old boy had complete amputations of his right thumb, index, and middle fingers and the distal phalanx of the ring finger. B, He underwent primary coverage with a groin flap. C, A left second toe-to-thumb transfer has provided excellent opposition and pinch of the new thumb. D, The donor site in his left foot is inconspicuous.



Fig. 51-4 A and B, This 9-year-old boy had an amputation of his left thumb at the midproximal phalanx level. C-E, The thumb was reconstructed with a left trimmed great toe transfer with excision of the medial portion of the proximal and distal phalanges of the great toe. F, The donor site in the left foot. G and H, The trimmed toe transfer has excellent function and appearance similar to that of a thumb.

A child with a hypoplastic or absent thumb but normal fingers should be considered for pollicization of the index finger. Toe transfers have been described for the treatment of isolated thumb hypoplasia⁵² but the benchmark is pollicization of the index finger.^{22,26,40-41,44,52-53} However, in congenital constriction ring syndrome in which the thumb is absent distal to the carpometacarpal joint, the proximal structures are normal, and the child will benefit more from a toe-to-thumb transfer (Fig. 51-5).

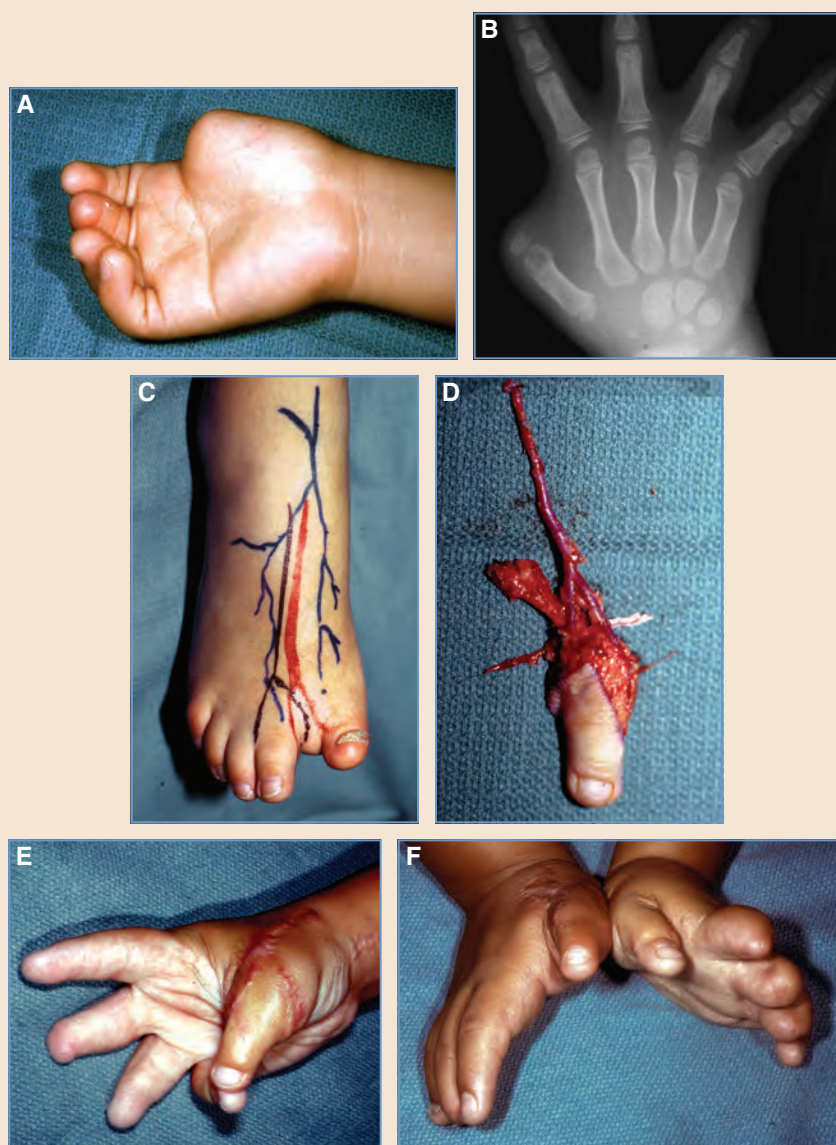


Fig. 51-5 A and B, This 3-year-old girl had congenital constriction ring syndrome affecting her right thumb, index finger, and middle finger. The thenar muscles were normal. C and D, She underwent a right second toe-to-thumb transfer. Her right great toe was abnormal, and she had a left above-knee amputation because of the congenital constriction ring syndrome. (The red line indicates the first dorsal metatarsal artery, the blue markings are the dorsal subcutaneous veins, and the black markings are the surgical incisions.) E and F, The postoperative functional result and a comparison of the appearance of the toe transfer with the contralateral thumb.

The choice of using a great toe or a second toe transfer should be individualized. Various toe transfers have been reported; however, the most appropriate one to transfer for each anomaly is controversial. Although a great toe transfer for thumb reconstruction may produce the most functional and aesthetically pleasing result (see Fig. 51-4), most parents choose the second toe for transfer in children.⁶² The donor site defect is much more obvious after a great toe transfer compared with the donor site after a second toe transfer. Because most parents have difficulty with their decision to “mutilate” a normal foot,²⁶ even if it means providing improved function to the hand, they tend to choose a second toe transfer because of the less obvious donor-site defect (see Figs. 51-2 and 51-3). Concerned families can be advised that parents and children are very accepting of the donor-site defect after second toe transfer, and this is true even as children enter adulthood.⁶³⁻⁶⁵ Patients prefer a second toe transfer in cultures in which thong-type sandals are worn, because the thong remains positioned between the great and third toes. After a great toe transfer, weight-bearing is shifted underneath the second and third metatarsal heads but does not necessitate modified footwear.⁵⁰ In general, parents can be reassured that sports activities are not significantly affected by second toe transfers; however, impairment in push-off and running speed and difficulties walking on uneven or sloped surfaces have been documented after great toe transfers.^{20,66-69}

Although it is technically possible to perform a toe transfer in patients younger than 1 year of age, most authors recommend waiting until they are 18 months of age, when the anesthetic risk is lower and the size of the digital vessels is somewhat larger.^{17,18,20} Children begin to use their hands during the first year of life; however, most authors agree that successful cortical reintegration can occur up to the age of 3 years and possibly even later.^{12,37,70} Waiting longer allows the family ample time to make an informed decision about the need for reconstructive surgery. Late toe transfers in children older than 10 years of age have been reported; however, even though the toe survives, children older than 8 years of age do not achieve normal sensibility in the transferred toe.⁷⁰ Parents should be counseled that the likelihood of cortical reintegration may be better if the procedure is performed when their child is younger.^{14,33,51,70}

Toe transfers have been used to reconstruct traumatic injuries in children and adolescents.⁵¹ Indications include thumb amputations (see Figs. 51-2 and 51-4), multiple finger amputations (see Fig. 51-3), metacarpal hand reconstruction, joint reconstruction, and pulp loss for insensibility.^{12,42,51} Toe transfers for trauma are usually performed later, after amputations have healed and parents have had time to decide, but an immediate toe transfer is very rarely considered for reconstruction of a degloved thumb with an intact bony skeleton and flexor and extensor tendons.

For microsurgical thumb reconstruction, second toes are used for children whose parents do not like or cannot tolerate loss of the great toe, for children with a second toe of reasonable size compared with that of the opposite thumb, and for children whose parents may be satisfied with less than optimal function and/or appearance of the thumb (see Figs. 51-2 and 51-3). Great toe transfers tend to provide a broader and stronger surface area for pinch compared with second toe transfers.⁶⁶ Complete great toe transfers and trimmed toe transfers (see Fig. 51-4) are more commonly used in children with traumatic thumb amputations. Great toe wraparound techniques should not be performed in children, because they limit the potential for growth.

Alternative options to lengthen digits in cases of partial transverse arrest include web space deepening, nonvascularized toe phalangeal bone grafting, and distraction lengthening. Augmentation of hypoplastic digits with nonvascularized toe phalangeal bone grafts may be used as an

adjunctive procedure in children with adactyly to provide improved length and possibly some growth potential when harvested extraperiosteally and performed before 1 year of age⁷¹⁻⁷⁵ (Figs. 51-6 and 51-7). However, toe phalangeal bone grafts cannot provide the improved motion, sensibility, length, and function provided by a toe transfer. In our experience, growth after nonvascularized toe phalangeal bone grafts has been modest at best.

Bone lengthening in the hand is an application of the established technique of distraction osteogenesis.⁷⁶ Lengthening the metacarpals in cases of symbrachydactyly provides increased length and a greater lever arm and depth of grasp, but it does not provide motion.¹⁸ The length gained with distraction osteogenesis is superior to that achieved by phalangeal bone grafting, although it requires a prolonged period of treatment.^{73,74,76} Because of the lack of motion obtained with distraction lengthening, toe transfer remains the optimal treatment option.



Fig. 51-6 A and B, This 4-year-old girl had a transverse failure of formation of her left middle and ring fingers at the level of the proximal interphalangeal joints. C, She underwent nonvascularized toe phalangeal bone grafts from the proximal phalanges of the second and third toes. D and E, Subsequent lengthening of the middle and ring fingers. Radiographs show the stabilization of the toe phalangeal bone grafts to the proximal phalanges using longitudinal K-wires. F, Subsequent shortening of the second and third toes in the donor left foot.

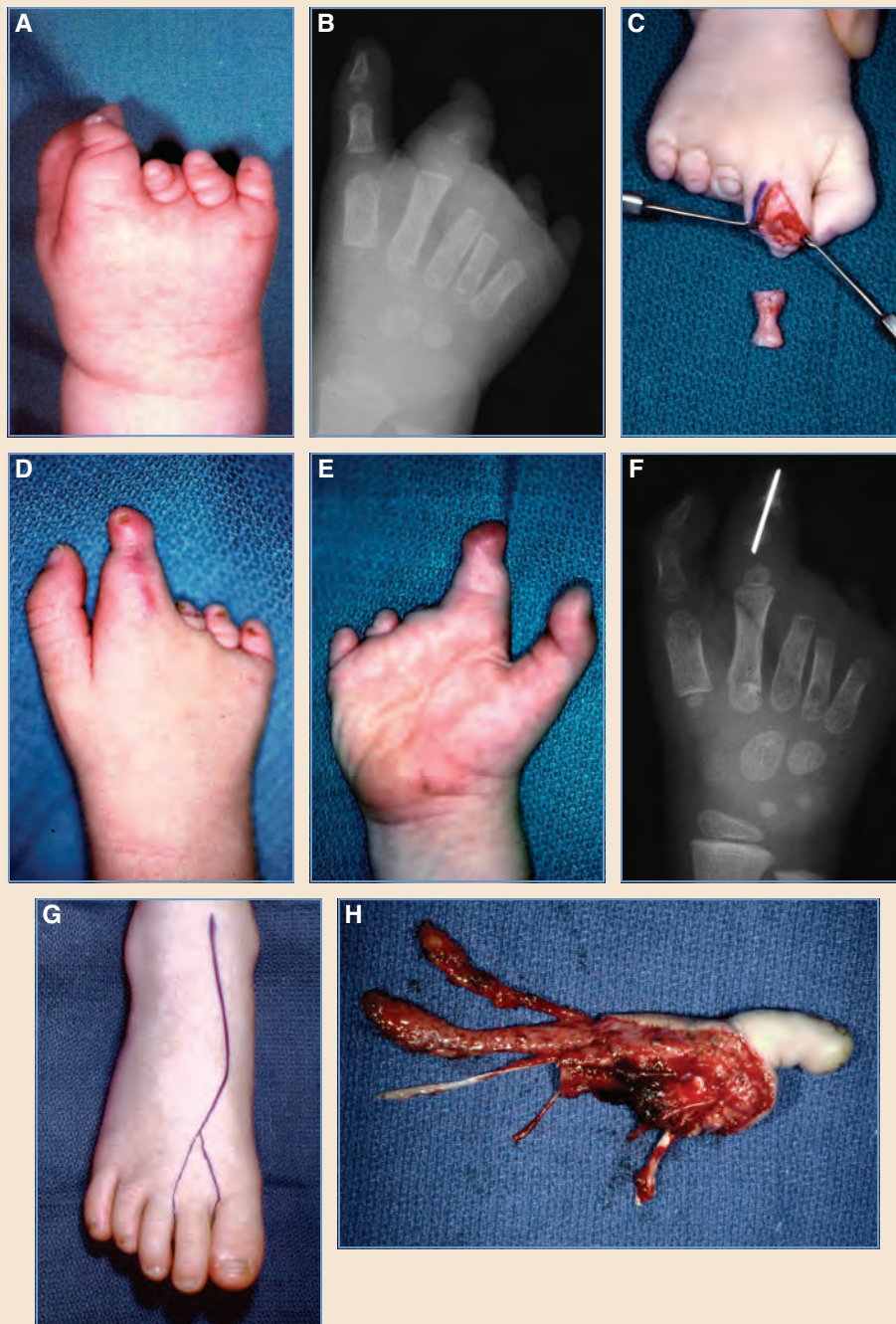


Fig. 51-7 A and B, A transverse failure of formation of the right hand with a remnant of the proximal phalanx of the index finger. C, The patient underwent nonvascularized toe phalangeal bone grafting of the right index finger at 6 months of age. D-F, Subsequent lengthening and stabilization of the right index finger 1 year postoperatively. G and H, At 2 years of age, he had a right second toe transfer into the middle finger position. (The blue marking indicates the surgical incisions.)

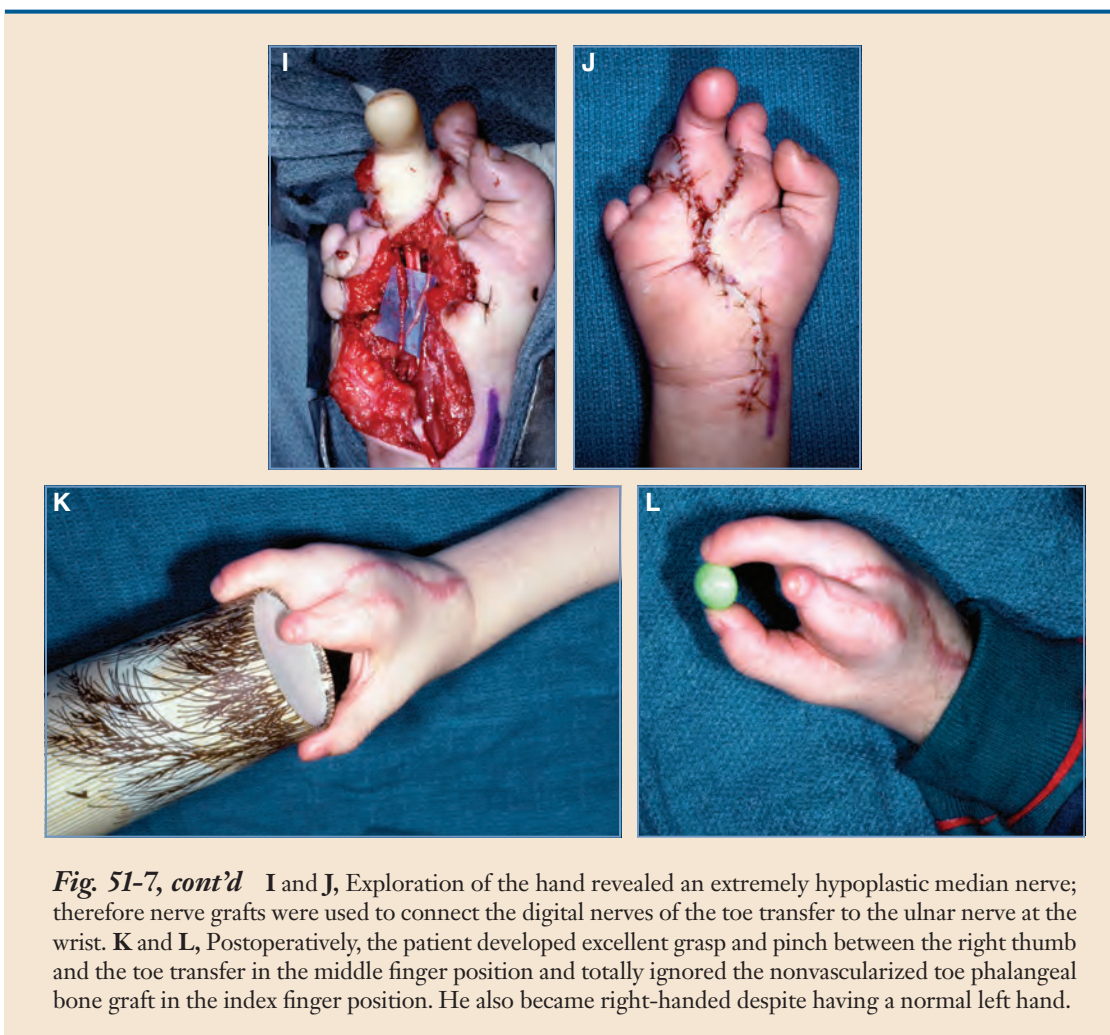


Fig. 51-7, cont'd I and J, Exploration of the hand revealed an extremely hypoplastic median nerve; therefore nerve grafts were used to connect the digital nerves of the toe transfer to the ulnar nerve at the wrist. K and L, Postoperatively, the patient developed excellent grasp and pinch between the right thumb and the toe transfer in the middle finger position and totally ignored the nonvascularized toe phalangeal bone graft in the index finger position. He also became right-handed despite having a normal left hand.

Contraindications

The most obvious contraindication to toe transfer is the absence or significant deformity of the toes. In general, however, even in children with severe cleft foot deformity, at least one of the two toes can be transferred without affecting the child's gait. Other contraindications include an inexperienced surgical team and/or surgical unit, absent recipient nerves, a transverse deficiency proximal to the distal carpus, and psychosocial factors in the parent or child.⁷

The family should be informed of and prepared psychologically for the significant amount of time and energy that will be required during the preoperative and postoperative period. The family must understand and accept that a toe transfer can fail. If this occurs, their child will have the same deficit in the hand and a new deformity in the foot. If the parents and/or the child cannot accept the time commitment and the potential risk of failure, this surgery should not be considered.

Toe transfer surgery is never urgent, and parents should be reassured that the operation can be performed when they are comfortable with it. Waiting until the child is 2 to 3 years of age or even older is perfectly reasonable, because the ability for central motor and sensory reintegration of the new digits is still possible.

PREOPERATIVE PLANNING

A child with one normal hand and a contralateral adactylous hand or a contralateral hand with a single digit or isolated thumb should be considered for a toe transfer (Figs. 51-8 through 51-11; see Box 51-1 and Fig. 59-7). Even in symbrachydactyly, in which some of the proximal anatomy may be anomalous or absent, a toe transfer can provide improved function. Toe transfers for unilateral adactyly should be positioned to allow the widest grip possible, because the opposite, normal hand will probably be used for all fine manipulation.



Fig. 51-8 A-C, This 3-year-old girl had a severe radial longitudinal deficiency affecting the left thumb, index finger, and middle finger. D, Dissection of the hand revealed normal extensor tendons and flexor tendons extending to the remnant of the base of the proximal phalanx of the thumb. E, Dissection of the right second toe transfer showing the extensor digitorum communis tendon and branches of the superficial and deep peroneal nerves. F and G, The postoperative result. A portion of the right second toe metatarsal was used to elongate the proximal phalanx of the left middle finger.



Fig. 51-9 A and B, This 6-month-old girl had a transverse failure of formation of all four fingers and an unstable thumb at the metacarpophalangeal joint. C, The parents refused a toe transfer. At 6 months of age the patient underwent an attempt at creating a post on the ulnar border of the hand using a nonvascularized toe phalangeal bone graft. D and E, The result of the nonvascularized toe phalangeal bone graft is shown, with minimal lengthening of the ulnar border of the hand. F and G, She subsequently underwent a left second toe transfer into the small finger position at 2 years of age. At 2 years and 7 years postoperatively, she had very precise pinch. H and I, At 2 years and 7 years postoperatively, she had strong grasp between the thumb and the toe transfer in the small finger position. The patient and her parents refused to allow the nubbins to be excised. J, The appearance of the left foot 7 years postoperatively.

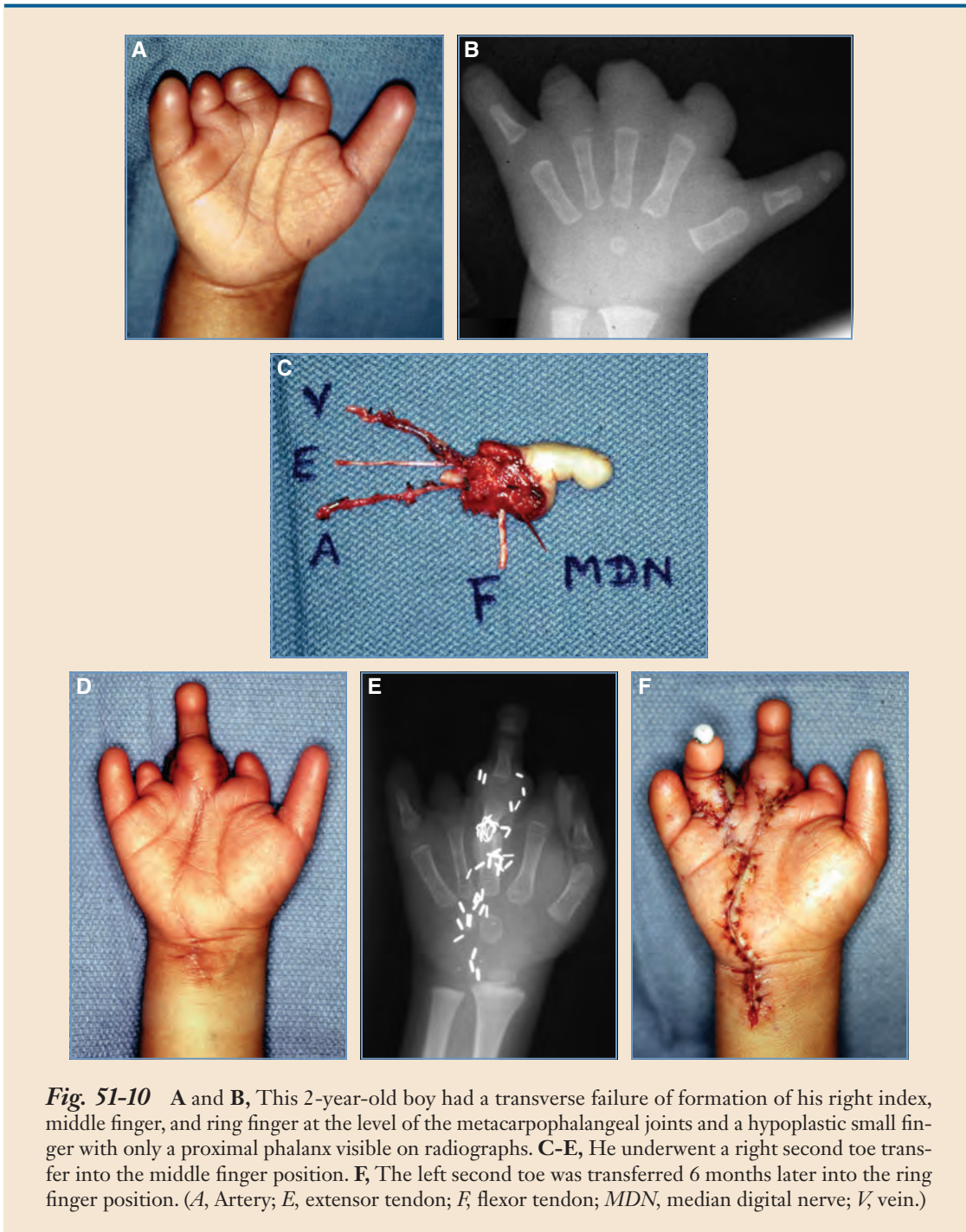




Fig. 51-10, cont'd G and H, The hand is shown 3 years postoperatively. I-K, Pinch function and grasp function and the appearance of both feet 3 years postoperatively.



Fig. 51-11 A and B, This 1-year-old boy had a transverse failure of formation of his left index, middle, ring, and small fingers at the level of the distal metacarpals, but his thumb was present. C-E, At 2 years of age, he underwent simultaneous bilateral second toe transfers into the middle and small finger positions of his left hand. F, The immediate postoperative result. G-I, The appearance of his left hand 3 years postoperatively. He is able to pick up small objects and use his left hand as an assist hand for grasping and lifting. (A, Artery; DN, digital nerve; E, extensor tendon; F, flexor tendon; N, dorsal nerve; V, vein.)

In children with bilateral deficiencies, the dominant hand should be reconstructed to allow opposition for fine motor control, whereas the opposite hand should have wider positioning for grasp.³ For a dominant hand, one toe should be placed in the thumb position to create a radial post and another toe placed in the middle or ring position to facilitate fine pinch. For a non-dominant hand, one toe is placed in the thumb position, and the other toe is placed in the ring or small finger position to provide wide grasp.

The primary requirements of a reconstructed thumb include stability, sensibility, the ability to oppose, and sufficient flexion and extension for grasping.^{20,66,77} Positioning a toe transfer depends on the presence of a competent thumb carpometacarpal joint. If this joint is absent, the transferred toes will have to be positioned so that they face each other, because most of the motion will occur at the interphalangeal joints. When transferring a great toe for thumb reconstruction, surgeons should consider the relative valgus angulation of the first metatarsophalangeal joint. Typically the first MTP joint has 10 degrees of valgus angulation; therefore transfer of the ipsilateral great toe to the thumb results in optimal positioning, with the new thumb pointing ulnarly.

Positioning and Marking

The child is placed in a supine position on the operating table, and all prominences are well padded. General anesthesia is given in all cases. Although an axillary block used in conjunction with general anesthesia has been shown to increase peripheral circulation to the hand, the technique is rarely applied in children.³⁵ Some surgeons have advocated axillary blocks to diminish the need for intraoperative and postoperative analgesia, whereas others have recommended continuous brachial plexus infusion postoperatively.^{8,20} Because of the length of the surgery, a Foley catheter is always placed to monitor the child's urine output and overall fluid status. Each patient is hydrated with standard intravenous fluids, but a central venous catheter is not used routinely.

Preoperatively, a handheld Doppler ultrasound probe is used to mark the location of the dorsalis pedis artery and/or the posterior tibial artery, depending on the local anatomy and the needs of the recipient hand.^{19,78} The dorsalis pedis artery is traced distally into the first dorsal metatarsal artery (FDMA), which lies between the great toe and second toe metatarsals. The location of the dorsal veins may be marked after a tourniquet is placed around the calf and before the start of the case, or after the patient is draped and the thigh tourniquet is inflated to 100 mm Hg (see Fig. 51-5, C). A tourniquet is applied around the upper arm. Because of the need for a split-thickness or full-thickness skin graft in most cases, a sterile thigh tourniquet is used to allow later access to the groin or thigh.

SURGICAL TECHNIQUE

If two teams are available, the hand and foot can be dissected simultaneously. Before beginning the operation, the teams convene to review the anatomy, the radiographs, and the needs of the particular child. If only one team is available, the hand usually is dissected first to confirm that the specific tendons and digital nerves are available for subsequent tendon and nerve repairs.

Dissection of the Foot

Dissection of the foot is performed under tourniquet control after exsanguination with an Esmarch bandage. Either 2.5× or 3.5× loupe magnification is sufficient for dissecting the structures in a child's foot. A racquet-shaped or V-shaped incision is marked on the foot, surrounding either the great or second toe (see Figs. 51-5, C, and 51-7, G). The dorsal skin incision begins proximally at the level of the cuneiform joints and extends distally in a longitudinal fashion over the dorsal

aspect of the first and second intermetatarsal space. This allows dissection of the dorsal veins and identification of the dorsalis pedis artery. The V-shaped dorsal skin flap extends either around the tibial aspect of the great toe and first web space or around the first and second web spaces, depending on whether the great toe or second toe is harvested. The two incisions usually create a plantar triangular flap with its apex at the MTP joint.

Dissection is continued through the dorsal subcutaneous tissues, with careful cautery of small draining veins. Usually, one dominant dorsal vein can be identified and dissected distally to the level of the dorsal V-shaped flap and proximally as far as the level of the ankle joint, if needed. The dorsalis pedis artery and deep peroneal nerve are identified proximally by dividing the extensor hallucis brevis tendon and dissecting from proximal to distal to identify the FDMA.⁷⁹ Alternatively, dissection of the FDMA can be started distally in the first web space, tracing the FDMA from distal to proximal into the dorsalis pedis artery, while an assistant retracts the great and second toes apart.^{7,12,80} Depending on which system appears more dominant, the dorsal or plantar arterial system is chosen for transfer.⁸ The dorsalis pedis artery typically divides into two branches at the level of the tarsometatarsal joint. The FDMA continues distally between the first and second metatarsals in one of three patterns, and the second communicating branch passes deep to anastomose with the plantar arch and continues distally as the first plantar metatarsal artery (FPMA).

The location and size of the FDMA can be quite variable.⁵⁷ It may lie in a dorsal, intramuscular, or plantar position. If the FDMA cannot be identified, the FPMA may be used for anastomosis and extended by a vein graft.³ If both the dorsalis pedis and the first dorsal metatarsal arteries are absent or hypoplastic, the great toe may still be transferred using the medial plantar arterial system.⁸¹ In most cases we use only one artery and one vein as the vascular pedicle.

In children with a congenital cleft foot, the great toe is usually transferred on the posterior tibial artery pedicle. The dissection begins proximally, just posterior to the medial malleolus, where the artery lies between the flexor digitorum longus and the flexor hallucis longus tendons, accompanied by the tibial nerve and posterior tibial vein. The artery can be traced distally to the medial plantar arterial system supplying the great toe.

The flexor hallucis longus tendon to the great toe or the flexor digitorum longus and flexor digitorum brevis tendons to the second toe can be identified by dissection through the plantar fascia and flexor tendon sheath at the level of the MTP joint. On either side of the tendon, the medial and lateral plantar digital nerves are identified and traced back to the level of the common digital nerves. Extra length may be gained by splitting the bifurcation of the nerve more proximally before transecting each digital nerve. The digital nerves are tagged with small microvascular clips to make their identification easier when it is time to repair them to the digital nerves in the hand.

The proper plantar digital arteries lie dorsal to the digital nerves and can be traced proximally to the FPMA. This vessel can be preserved and used as a secondary recipient artery if needed. All branches from the FDMA and/or FPMA supplying the second toe should be cauterized or clipped if the great toe is transferred. Alternatively, all branches from the FDMA or FPMA supplying the great toe should be cauterized or clipped if the second toe is transferred. The dissection is continued in the first web space, identifying the deep peroneal nerve, which must be carefully divided into separate fascicles supplying the adjacent great toe and second toes. The fascicles serving the toe to be transferred are then divided from the main trunk of the deep peroneal nerve for later repair to the superficial branch of the radial nerve in the hand.

The extensor hallucis longus is dissected proximally and divided when the great toe is transferred. Alternatively, the extensor digitorum communis and extensor digitorum brevis tendons are dissected and divided proximally when the second toe is transferred (see Fig. 51-8, *E*). Both the

flexor and extensor tendons should be divided as long as possible to allow repair away from the site of osteosynthesis and to provide enough length in cases in which the proximal motor units are short or deficient. The flexor hallucis longus to the great toe or the flexor digitorum longus to the second toe may be difficult to harvest without creating a long incision on the plantar surface of the foot. To gain more length, the tendon may be divided through a dorsal approach after the metatarsal osteotomy.

When the thumb is reconstructed using a second toe, the osteotomy may be performed either at the MTP joint or more proximally in the metatarsal shaft. The great toe may be harvested at the level of the MTP joint or through the base of the proximal phalanx; but a more proximal osteotomy through the great toe metatarsal may interfere with gait. If the MTP joint is chosen, the osteotomy is performed at an oblique angle so that the joint is fixed in hyperextension to prevent further hyperextension of the toe when it is placed in the thumb position.

After the osteotomy has been performed, further dissection of the arterial and venous pedicles to increase their length can be completed. All small branches are carefully clamped with small microvascular clips or cauterized with bipolar electrocautery. Once the dissection is complete, the tourniquet is deflated to allow reperfusion of the toe, which may take up to 15 minutes. The vascular pedicle can be irrigated with dilute (1:20) papaverine solution, and laparotomy pads soaked in warm irrigation fluid may be placed over the foot to improve digital blood flow.

Dissection of the Hand

If two teams are available, dissection of the hand and the foot may be performed simultaneously. In symbrachydactyly a horizontal incision may be made across the residual nubbins to expose tiny vessels that can be traced proximally.¹⁹ The vascular, nerve, and tendon anatomy is usually anomalous in symbrachydactyly; in these cases, the hand is dissected before the foot to ensure that adequate nerves and tendons are present in the hand.⁴⁹ In congenital constriction ring syndrome, normal anatomy is usually preserved out to the level of the amputation³ (see Fig. 51-8, D).

The dorsum of the hand is incised to expose the extensor tendons and, if available, the superficial branch of the radial nerve. The incision is usually designed longitudinally so that the V-shaped dorsal skin flap from the toe will interdigitate after transfer of the digit. A dorsal vein, preferably a branch of the cephalic or basilic vein, is identified and tagged for the later venous anastomosis.

The volar surface of the hand is opened in a zigzag fashion across the wrist crease. The median and/or ulnar nerve are carefully dissected to assess potential sites for coaptation of the plantar digital nerves. Through this same incision, the flexor tendons are dissected and assessed for their excursion. The transverse carpal ligament should be left intact. Once adequate tendons, nerves, and vessels have been identified in the hand, the wounds are provisionally closed with staples to prevent excessive swelling. The tourniquet on the upper extremity is deflated, and the vascular pedicle to the toe transfer is subsequently divided. The arterial pedicle is usually ligated with 3-0 silk and reinforced with small microvascular clips.

Closure of the Foot

The defect in the foot can be closed primarily; rarely is a split-thickness skin graft required. The defect left after a second toe transfer is closed by first closing the gap between the first and third metatarsals. The intermetatarsal ligaments arising from the adjacent metatarsals are sutured together using a heavy Ethibond suture, closing the gap created by harvesting the toe. Meticulous hemostasis is obtained with Bovie electrocautery. If drainage is persistent, a small Jackson-Pratt drain can be left in the wound after closure. The skin is closed with interrupted 4-0 chromic catgut horizontal mattress sutures.

The defect created by harvesting a great toe is similarly closed primarily. Reconstruction of an intermetatarsal ligament is not required.

Transfer of the Toe

After a toe is harvested, the recipient site in the hand is prepared for osteosynthesis. The tourniquet is reinflated to 250 mm Hg to ensure a bloodless field. If a segment of metatarsal has been harvested, the periosteum is elevated from its base, and the periosteum is elevated from the distal aspect of the recipient metacarpal bone in the hand. Rotation of the toe transfer is determined by placing the base of the metatarsal at the osteosynthesis site and checking the relationship of the toe to the remaining digits or to the proposed location of a future toe transfer. In general, a toe-to-thumb transfer is placed in 120 degrees of pronation but should be adjusted as needed to allow adequate grasp and/or pinch, depending on the needs of the child. The exact rotation of the toe is marked on the corresponding surfaces of the metatarsal and metacarpal at the osteosynthesis site to allow fixation in the correct position. Any excess length of metatarsal can be removed using an oscillating saw.

Excellent fixation can be obtained with the use of 90-90 intraosseous wiring between the base of the metatarsal and the recipient bone in the hand. With the use of a 0.035-inch K-wire, two holes are drilled perpendicular to each other, approximately 5 mm from the base of the metatarsal and parallel to the cut surface of the bone. Similarly, two matching holes are drilled through the metacarpal. The surgeon passes two 26-gauge stainless steel wires through the holes and twists them clockwise while pulling perpendicular to the osteosynthesis site. Alternating back and forth between the two wire loops allows the bone ends to be opposed accurately (see Figs. 51-4, *E* and 51-10, *E*). The surfaces of the stainless steel wires change from a glistening, shiny appearance to a dull gray when the wire has reached its maximal tension. K-wires may be required for added stability but are not usually necessary. Some surgeons use K-wires exclusively, arguing that they do less damage to the growth plate, are quicker to apply, and are stable enough alone to achieve union.⁷

If the toe has been harvested through the MTP joint, the base of the toe proximal phalanx creates a new joint with the head of a metacarpal, but stability and mobility may be difficult to achieve because of the limitations of collateral ligament repair. The strong volar plate can be repaired to the volar aspect of the metacarpal using a heavy suture or mini-suture anchors.

In situations in which a suitable foundation for a thumb is absent, the adjacent index finger metacarpal may be pollicized. This allows osteosynthesis of the base of the toe transfer to the pollicized index metacarpal base. Alternatively, a second toe, with a long metatarsal, may be stabilized with K-wires or interosseous wires to the radius (Vilkki procedure) or with a residual carpal bone to create a pseudarthrosis.⁵⁰

Once bony stability has been achieved, the extensor tendon is reconstructed. The extensor hallucis longus to the great toe or the extensor digitorum communis to the second toe is repaired to an available extensor tendon in the hand. The repair is performed under tension with overlap of the tendons. Multiple horizontal mattress sutures using 4-0 nylon are sufficient to allow early motion. When reconstructing a new thumb, the extensor indicis proprius or an adjacent extensor tendon (extensor digitorum communis) to the index finger remnant is usually transferred as a donor tendon. When a finger is reconstructed, a quadrigia effect caused by tethering of the extensor digitorum communis with a common muscle belly to the absent digits must be prevented by releasing adjacent extensor tendons.

Only one flexor tendon, usually the flexor hallucis longus to the great toe or the flexor digitorum profundus to the second toe, is repaired to a recipient flexor tendon in the hand, especially

in children with symbrachydactyly or a transverse deficiency. If a flexor tendon with adequate excursion cannot be identified, a primary tendon transfer may be required. The flexor tendon repair is performed by overlapping or weaving the tendons using multiple 4-0 nylon horizontal mattress sutures. To prevent a quadrigia effect, adjacent flexor tendons that may tether the newly reconstructed flexor tendon to the toe are divided.

Neurorrhaphy of the medial and lateral digital nerves and the deep peroneal nerve of the toe transfer to the recipient nerves in the hand is then performed under a microscope. The site of neurorrhaphy is determined by the quality of recipient nerves in the hand and the length of the nerve harvested from the foot. If possible, nerves are repaired more proximally to ensure the best recipient nerve is selected. The medial and lateral digital nerves are repaired in an end-to-end epineurial fashion to residual stumps of the proper or common digital nerves using 10-0 nylon in an interrupted suture technique. The deep peroneal nerve is repaired to a branch of the superficial radial nerve. If recipient digital nerves are inadequate or unavailable, the toe digital nerves can be coapted in an end-to-side fashion to the median or ulnar nerves, or, alternatively, nerve grafts from the superficial or deep peroneal nerves can be interposed (see Fig. 51-7, *I*).

Vascular anastomoses should be performed after the tourniquet has been deflated to allow reperfusion of the upper extremity and then reinflated after 20 minutes. Before reinflation of the tourniquet, a microvascular clamp can be applied to the recipient artery (radial or ulnar artery) to be sure the other digits remain perfused. If the hand remains pink, the arterial anastomosis can be performed in an end-to-end fashion.

The venous anastomosis is usually performed before the arterial anastomosis. It is preferable to harvest as long a vein as possible and anastomose it end to end to a large, more proximal vein, typically the cephalic or basilic vein at the wrist. The anastomosis is performed using standard microsurgical techniques under a microscope with 10-0 nylon sutures. After the vein has been anastomosed, the recipient vein is occluded proximally with a small microvascular clamp.

The site of the arterial anastomosis depends on which toe arterial system (dorsal or plantar) was harvested and the location of the recipient radial or ulnar artery. End-to-side or end-to-end anastomosis to the ulnar artery or the dorsal branch of the radial artery in the anatomic snuffbox can be performed if a long arterial pedicle is available, usually the dorsalis pedis–FDMA or the posterior tibial artery. If the FPMA system was harvested, it is usually too short to reach the radial artery. Occasionally, it can be anastomosed to the superficial palmar arch or to a common digital artery, but it is usually better to elongate the FPMA with an interposition vein graft anastomosed on a back table before performing bony osteosynthesis and tendon and nerve repairs. The arterial anastomosis is performed similar to that of the vein, using 10-0 interrupted nylon sutures.

The tourniquet is deflated, leaving the single microvascular clamp on the proximal side of the venous anastomosis in place. This creates a high-pressure column of blood running through the toe, theoretically expanding collapsed vessels, which may be in spasm. The vascular anastomoses are bathed in dilute (1:20) papaverine solution, and the hand is warmed with laparotomy pads soaked in warm irrigation fluid. The patency of the anastomoses is checked by the milking test, and the vascular clamp is removed from the proximal side of the venous anastomosis.

The incisions are loosely closed with interrupted 4-0 and 5-0 chromic catgut mattress sutures. If the closure is under too much tension, a split-thickness skin graft should be applied. A 14/1000-inch split-thickness graft from the thigh is preferable. The skin graft is not meshed, but small fenestrations are made with a No. 11 blade scalpel to allow drainage. Alternatively, a full-thickness skin graft from the groin is used. The skin graft can be excised at a later date to improve appearance. A sterile pulse oximeter probe is applied to the toe transfer before the dressings are applied and is left in place for postoperative monitoring.¹⁶ A loose sugar-tong splint is applied to the upper extremity, and the foot is immobilized in a posterior plaster splint.

POSTOPERATIVE CARE

The child is monitored in a pediatric intensive care unit for 1 to 2 days postoperatively and then transferred to the general pediatric floor for another 5 to 7 days. The hand and foot are elevated, and the hand is kept warm with a heating blanket. The core body temperature, heart rate, blood pressure, respiratory rate, systemic oxygen saturation and urine output are monitored closely. The toe transfer is monitored hourly for color, capillary refill, oxygen saturation, and pulse rate. Differential pulse oximetry is superior to temperature monitoring and is a simple, continuous way to noninvasively monitor toe-to-hand transfers in children.³⁶ The toe transfer pulse rate corresponds with patency of the arterial anastomosis, and any decrease or loss of the pulse rate compared with the systemic heart rate may indicate that the arterial anastomosis is compromised. The oxygen saturation corresponds to the patency of the venous anastomosis, and any decrease in the oxygen saturation for an extended period of time, compared with the systemic oxygen saturation, may indicate thrombosis of the venous anastomosis. If the appearance of the toe transfer or the differential pulse oximetry indicates compromised perfusion to the toe transfer, the patient should be immediately returned to the operating room for exploration and possible revision of the microsurgical anastomoses.

The child is kept sedated with the use of opiates and benzodiazepines, as needed. Anticoagulation with dextran 40 (0.36 ml/kg/hr) is continued for 5 days. An 81 mg aspirin tablet is started on postoperative day 1 and continued daily for 4 weeks. The dextran infusion rate is reduced to half on postoperative day 6 and then discontinued on day 7. A first-generation cephalosporin is given intravenously for 48 hours postoperatively. Intravenous fluids are maintained based on weight until the child is tolerating an oral diet. The hand and foot dressings are changed on postoperative day 5 to 7. If there is any concern with the perfusion to the toe transfer, the cast and dressings should be released or removed completely, because occasionally the plaster splint and dried blood can combine to form a constrictive cast, producing external pressure on the anastomoses. Ambulation on the donor foot can begin as soon as the child is comfortable doing so.

The plaster hand and foot splints are removed at the first postoperative visit at 2 weeks, and the toe transfer is inspected for capillary refill and skin graft take. The foot incision is checked; occasionally, some patients require local wound care for minor dehiscence or oral antibiotics for localized cellulitis. A removable Thermoplast splint is fabricated to protect the toe transfer, and the parents and child are instructed on passive and active range-of-motion exercises. A hand therapist may assist with the therapy if the family is apprehensive. K-wires are removed if necessary at 6 weeks. The parents may relate that their child is having dysesthesias as reinnervation occurs. They should be instructed that these sensations are normal and will resolve as the nerves regenerate.⁷

TREATMENT OUTCOMES AND COMPLICATIONS

Toe-to-hand transfers have helped restore function in children with congenital differences of the hand, including longitudinal deficiency, transverse deficiency, symbrachydactyly, congenital constriction ring syndrome, and after severe trauma.

O'Brien et al²² reported the first microsurgical toe-to-hand transfer in a child in 1978. The great toe was successfully transferred to treat two children with thumb hypoplasia. In both children the toe transfer was being used for “many tasks” 1 year postoperatively, and no problems with gait were noted in either child. Nyarady et al⁴⁰ performed a second toe transfer for thumb hypoplasia in a 13-year-old boy in 1983. The patient developed 6 mm of two-point discrimination by 4 months after the operation and had satisfactory flexion and extension of the thumb. Although index finger pollicization remains the benchmark treatment for thumb hypoplasia and aplasia, these early reports fueled interest in treating children with more severe congenital hand differences with toe transfers.

Yoshimura⁸² reported the first large series of pediatric toe transfers in 1980. He successfully performed 33 toe transfers in 28 patients, 12 of whom were children. All but two cases were performed for traumatic amputations; one child had a congenital deficiency of the thumb, the index finger, and the middle finger, and another child had an intrauterine amputation of one finger. In all the pediatric cases, the epiphyseal plate was undisturbed, and growth potential was theoretically maintained. But based on a follow-up study 4 years after the initial paper, the growth of the transferred toes was found to be less than that of the contralateral toes.⁸³

In contrast, Chang and Jones²⁶ documented excellent potential for digital growth after toe transfer. A comprehensive radiographic study of toe transfers performed in children 2.8 to 13 years of age demonstrated that the epiphyseal plates remained open in 27 of 28 phalanges at a mean of 12 months' follow-up (range 1 to 36 months). In four patients in whom radiographic comparison was possible, the amount of linear growth of the toe transfer was equal to the growth of the contralateral, nontransferred toe. Wei and Mardini¹² found that the trimmed toe transfer does not affect growth, and they regularly use this technique in children with traumatic amputations. Kay and Wiberg⁸ found that the length of the toe transfer varied from 70% to 104% of the length of the contralateral toe.

May et al⁷⁷ reported on a case of bilateral toe-to-thumb transfers in a 9-year-old boy with aplasia of the thumb and the index and middle fingers of both hands. In each procedure the great toe was harvested because of a cleft foot anomaly. Eighteen months postoperatively the child required bilateral thumb interphalangeal joint arthrodeses for instability and an abductor digiti minimi transfer for improved thumb opposition. The patient had no gait abnormalities at the final follow-up and was using the thumbs regularly for many activities but continued to use the ring and small fingers for lateral pinch of small objects. Two-point discrimination was 9 to 10 mm at the 3-year follow-up visit.

Gilbert¹⁵ reported on 21 second toe transfers performed between 1977 and 1980 in 17 children with congenital hand differences. Four thumbs and 17 fingers were reconstructed in children ranging in age from 18 months to 13 years (12 transfers were performed in children younger than 3 years of age). One venous thrombosis occurred, which was detected 4 hours postoperatively and treated by immediate reoperation. The child was given 50 mg of aspirin daily for 10 days, and the toe transfer remained viable. Gilbert found that the results were better in children with congenital constriction ring syndrome, compared with results in children with congenital aplasia, because the structures were more "normal" proximally. Restoration of motion was minimal, with a mean active combined flexion of the proximal and distal interphalangeal joints of 40 degrees (range 0 to 80 degrees). Five secondary tenolysis procedures provided negligible improvement. Sensibility was assessed subjectively, and most children were unable to discriminate any difference between the transferred toe and the normal digits. Growth of the toe transfer was similar to that of the contralateral, nontransferred second toe. No deficits in gait were noted, but the dorsal donor site scar was sometimes hypertrophic.

Lister¹⁷ reported on his experience with 12 children 10 months to 8 years of age in whom the second toe was used to reconstruct the thumb in congenital constriction ring syndrome, symbrachydactyly, and true transverse arrest. Active motion was achieved in only 3 of the 11 children, and 1 child underwent an unsuccessful tenolysis 2 years postoperatively. Two-point discrimination was not assessed, but all children demonstrated "sweating and good tactile adhesion" by 6 months postoperatively.

Shvedovchenko²³ reported on 103 successful toe transfers in 66 children 3 to 14 years of age with congenital and posttraumatic deformities of the hand. The second toe, the small toe, and a composite of the second and third toes were used to reconstruct the hands of children with brachydactyly, ectrodactyly, adactyly, and traumatic loss. Functional outcomes and complications were not reported.

Vilkki¹⁹ reported his results of 18 toe transfers in children for congenital adactyly. One failure occurred, resulting from anesthesia complications. Five reexplorations for immediate postoperative vascular problems were required in the other 17 children. Pinch was restored in 14 of 17 hands. Linear growth of the transferred toes occurred without problems in the phalangeal epiphyses; however, the metatarsal head epiphyses showed evidence of growth arrest in two children.

Kay⁷ successfully transferred 125 toes in children without a single loss. In 1996 he published his initial series of 40 children (9 months to 14 years of age) with 66 toe transfers.^{8,9} A single toe was transferred in 14 children, and two toes were transferred in 26 children to reconstruct hands with congenital malformations, traumatic loss, or infection. Five children required early postoperative vascular reexploration, but infection occurred in only one case. At a mean of 45 months postoperatively, the children and their parents were reviewed for functional and psychological performance. Similar to earlier reports, the range of motion achieved was less than expected, with an average of 60 degrees total active range of motion. Tenolysis was performed in 15 children, and tendon transfers were required in 7. A secondary rotational osteotomy was performed in 4 children, and digits were augmented with nonvascularized phalangeal bone grafts in 3. In children in whom two-point discrimination was thought to be reliable, the mean was 5 mm, and all children developed protective sensibility.

Bellew and colleagues^{63,64} performed a detailed psychological evaluation of a subset of Kay's series 1 year and 10 years after transfer. One year postoperatively, 92% of parents thought the reconstruction improved their child's hand function, and 83% thought it improved the overall appearance of the hand. Ten years after transfer, 90% of parents and 88% of patients were satisfied with the outcome of the surgery. With regard to function, 90% of parents and 94% of patients stated the surgery had improved hand function. Patients who had grown into adulthood had scores within the normal range on depression and anxiety measures and a higher than average self-esteem.

Toe transfers have also been used for reconstructing congenital digital loss in older children. Spokevicius and Razdevicius¹⁴ successfully transferred four toes in three children 11 to 17 years of age with congenital malformations. The preoperative indication was improved appearance in all three children. One 17-year-old girl required reexploration and revision of the arterial anastomosis in the early postoperative period, and a 12-year-old girl developed arterial insufficiency on the first postoperative day, requiring heparinization and a brachial plexus block to restore flow. Two-point discrimination was documented in three of the four toe transfers, and grip strength and dexterity improved in all cases.

Wei and colleagues^{12,51} performed 45 complete toe or partial toe transfers in 28 children and adolescents (range 2 to 16 years of age) for traumatic defects. Three patients required postoperative reexploration. One toe transfer failed completely, and partial pulp loss occurred in two digits. Static two-point discrimination averaged 5 mm, and the range of motion averaged 69 degrees at the metacarpophalangeal joint, 38 degrees at the proximal interphalangeal joint, and 13 degrees at the distal interphalangeal joint of the second toe transfers. The average motion at the interphalangeal joint of the transferred great toes was 15 degrees.

Van Holder et al³⁸ reconstructed the hands of 14 children with congenital constriction ring syndrome, symbrachydactyly, and transverse failure of formation using staged, double second toe transfers. Postoperatively, all of the toes had sensibility and mobility and provided improved function and appearance; however, all patients required at least one secondary procedure.

I (N.F.J.) have performed more than 100 pediatric toe-to-hand transfers. In a published series of 82 transfers in 68 children with congenital hand differences, the overall success rate was 97.6%.⁴⁶ Seven toes (8.5% of transfers) required reexploration for salvage, and all children regained light touch perception and improved hand function. Jones and Kaplan⁴⁸ evaluated

15 children using a validated patient reported outcomes measure for the pediatric orthopedic patient population. They found that for most functional dimensions, children who underwent toe-to-hand transfer did not differ significantly from the “normal” pediatric population. Adolescent patients and their parents reported lower upper extremity function and sports/physical function scores relative to “normal” children. However, the same adolescent patients reported a higher overall happiness, compared with the general population.

CONCLUSION

Toe-to-hand transfer in children has become an accepted tool in the armamentarium of pediatric hand surgeons, although it remains a technically challenging undertaking, requiring a high level of skill on the part of the surgeon and the trust and patience of the child and parents. Miraculous improvement in function can be achieved in children affected by severe congenital hand differences such as symbrachydactyly, congenital constriction ring syndrome, and longitudinal deficiencies, and after traumatic amputations.

The appropriate age for toe transfer is 2 to 3 years of age, although the procedure can be successful in older children. Preoperative counseling providing the parents with the opportunity to meet and/or speak with other children and/or families who have undergone a toe transfer is very helpful. The child should be examined by a pediatrician for medical clearance and to rule out other associated anomalies before surgery. The appropriate toe to be transferred and the position to place the transfer on the hand are details that are specific to each child and should be discussed in detail with the child and parents before the operation. Informing the parents of the potential for the procedure to fail, with subsequent loss of the toe transfer, is essential.

A toe transfer involves many critical steps, but identification of the vascular pedicle and dissection of the toe are the most challenging. Positioning of the toe, osteosynthesis, and tendon repair are important steps that can be perfected with patience and experience.

Typically, only one artery and one vein are needed to provide adequate perfusion of the toe transfer. Closure of the incisions should be tension free, and split-thickness skin grafts may be necessary.

Postoperative care is critical to the success of a toe transfer. An initial stay in an ICU setting with sedation and transfer to a general pediatric floor after 2 to 3 days allows the most intensive monitoring during this critical period. Continuous differential pulse oximetry monitoring, intravenous dextran anticoagulation for at least 5 days, intravenous antibiotics, intravenous hydration, and a dressing change before discharge home help to ensure the success of a toe transfer.

The parents should be informed that sensation may take 3 to 6 months to return, and that motion of the transferred toe may improve over a 1- to 2-year period. These children may require a tenolysis, tendon transfers, bone grafting, an arthrodesis, an osteotomy, or scar revision to achieve their optimal functional and aesthetic outcome.

Although toe-to-hand transfer may not provide the most aesthetically pleasing result, and the motion achieved may seem less than ideal, the changes that occur in the lives of children after toe transfer are truly phenomenal. A 5-year-old child who is able to write his or her name for the first time, or an 8-year-old who can participate in sports such as baseball and gymnastics, provide the surgeon with far more feedback than the most sophisticated outcome instruments. In addition, simple activities of daily living such as bathing, dressing, and eating are made easier for most children, which greatly improves their self-esteem and is easily demonstrated in postoperative visits. A toe-to-hand transfer is the best procedure for reconstruction for children who have multiple missing digits resulting from traumatic amputations or congenital differences. This complex procedure will continue to be refined, but in the future may be supplanted by digital transplantation or by digital regeneration with genetically engineered tissues.

KEY POINTS

- Toe-to-hand transfer in children may be indicated after a traumatic injury or for congenital constriction ring syndrome, transverse deficiencies, cleft hand, and symbrachydactyly.
- Thumb hypoplasia in radial ray deficiency is best managed by index pollicization.
- Attention to detail is paramount in history-taking, clinical examination of the hand and foot, digital positioning, bony fixation, and postoperative support.
- Preparation of the toe and its vascular pedicle is the most challenging technical feature.

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52

Hand and Upper Extremity Trauma

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In children, the upper extremity—the hand in particular—is among the most commonly injured body parts, accounting for a significant percentage of emergency department visits.¹⁻³ Because the anatomy of the upper extremity is complex and the consequences of injury are potentially serious, proper diagnosis and timely treatment are essential. Most hand injuries are minor; however, some may be limb-threatening, even life-threatening. Toddlers have a propensity for fingertip injuries, especially crush injuries in closing doors, whereas older children and teenagers have sports-related hand injuries that are often closed fractures.⁴

Care of pediatric hand injuries poses particular difficulties and pitfalls in that children are unreliable historians and often do not cooperate with the hand examination or postoperative rehabilitation. Furthermore, their developing growth plates, cartilaginous caps, and growing bones require a surgeon who is thoroughly familiar with this unique anatomy, because these sites are frequently traumatized. Fortunately, children have remarkable healing capabilities and usually recover without complications and stiffness.

ANATOMY

The anatomy of a child's hand differs from that of an adult. The differences are typical to each system and change with the age of the child.

Skin

The skin of the hand, as in an adult, includes dorsal, elastic skin and palmar, glabrous, specialized skin. The palmar skin is thicker and is connected by fascial septa to the palmar fascia, allowing traction and durability during function.

The dorsal skin is more elastic in children than in adults, allowing more swelling to occur. In infants, a larger amount of subcutaneous fat may obscure some of this swelling and some of the bony landmarks on the dorsum of the hand.

Vascular System

The vascular supply of the upper extremity originates from the axillary artery, which becomes the brachial artery distal to the axilla (Fig. 52-1). The brachial artery courses along the medial intermuscular septum and enters the antecubital fossa medial to the bicipital aponeurosis. The major branches of the brachial artery are the profunda brachii, the superior and inferior ulnar collateral arteries, and the biceps myocutaneous arteries. The artery bifurcates into the radial and ulnar arteries in the proximal forearm approximately 1 cm distal to the elbow joint.

The radial artery courses laterally over the insertion of the biceps tendon, then over the fascia of the flexor digitorum superficialis and distally between the brachioradialis tendon and flexor carpi radialis. It then crosses the snuffbox, giving off a branch that dives between the two heads of the first dorsal interosseous muscle; it is the main contributor to the deep palmar arch and also forms the princeps pollicis.

The ulnar artery courses medially in the forearm deep to the median nerve and the two heads of the flexor digitorum profundus and flexor carpi ulnaris. The artery gives off the recurrent branches at the level of the flexor digitorum superficialis origin and the common interosseous artery deep to the pronator teres. This artery later divides into the anterior and posterior interosseous arteries.

Together with the ulnar nerve, the ulnar artery enters the hand through the Guyon canal. The artery then divides and gives branches to the deep and superficial palmar arches. The superficial palmar arch is mainly supplied by and is the continuation of the ulnar artery. The arch gives off digital branches to the thumb (the first web space), the common digital branches to the three web spaces, and one branch to the ulnar side of the little finger. The common digital branches divide into radial and ulnar digital branches to the fingers. Although the vascular structures are superficial to the nerves in the palm, they become deep to the nerves in the fingers.

Nervous System

The brachial plexus gives rise to the nerves in the upper extremity (Fig. 52-2). The plexus emerges from roots C5-T2 and the first thoracic root. Occasionally, C4 and T2 contribute to the brachial plexus. The rootlets merge to form the upper, middle, and lower nerve trunks and travel in close association with the subclavian vessels. The posterior cord of the brachial plexus gives rise to the axillary nerve and the radial nerve. Just distal to the clavicle, the trunks intermingle to form the posterior, lateral, and medial cords. More distally, these structures divide to form the major peripheral nerves of the upper extremity.

The posterior cord gives rise to the axillary and radial nerves. The lateral cord gives rise to the median nerve and the myocutaneous nerve and contributes to the median nerve. The medial cord forms the ulnar nerve and also contributes to the median nerve. The superficial location of the median, ulnar, and myocutaneous nerves places them in susceptible positions for penetrating injuries, whereas the axillary and radial nerves are more commonly injured during shoulder dislocation.

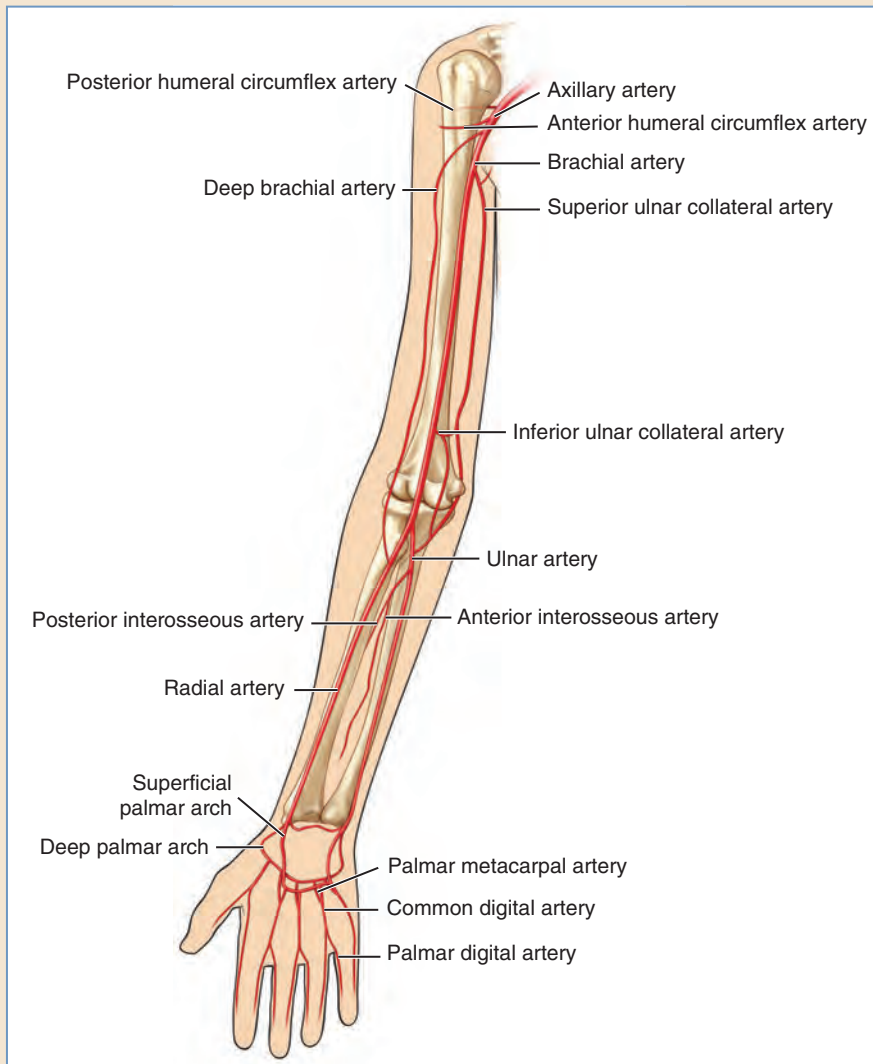
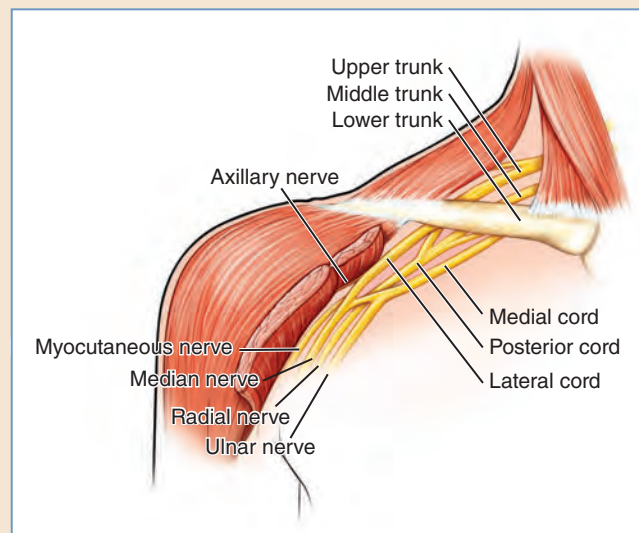


Fig. 52-1 Arterial anatomy of the upper extremity.

Fig. 52-2 The brachial plexus.



The axillary nerve provides motor innervation to the deltoid muscle and sensory innervation to the lateral and dorsal aspects of the shoulder. The myocutaneous nerve innervates the brachialis, coracobrachialis, and biceps muscles. It becomes the lateral cutaneous nerve of the arm and forearm, supplying sensation to the proximal radial portion of the forearm.

The median nerve travels with the brachial artery in the arm (Fig. 52-3). The median nerve crosses the elbow in the cubital fossa, medial to the brachial artery. It then enters the forearm through the two heads of the pronator teres. The median nerve innervates the flexors of the wrist and fingers. It courses on the inferior surface of the flexor digitorum superficialis and innervates the pronator teres, flexor digitorum superficialis, palmaris longus, and flexor carpi radialis. The anterior interosseous branch innervates the flexor pollicis longus, pronator quadratus, and the two radial muscle bellies of the flexor digitorum profundus.

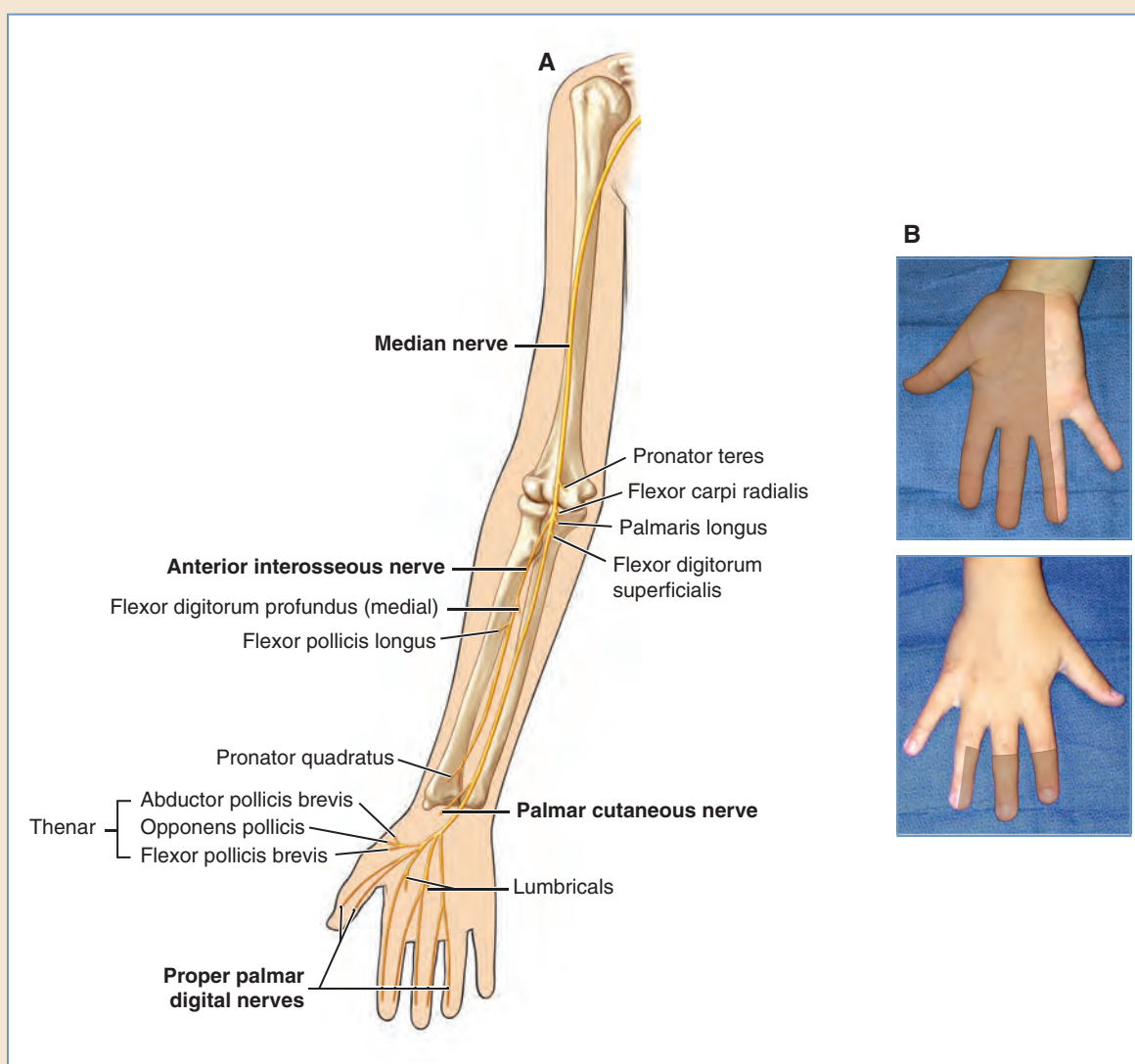
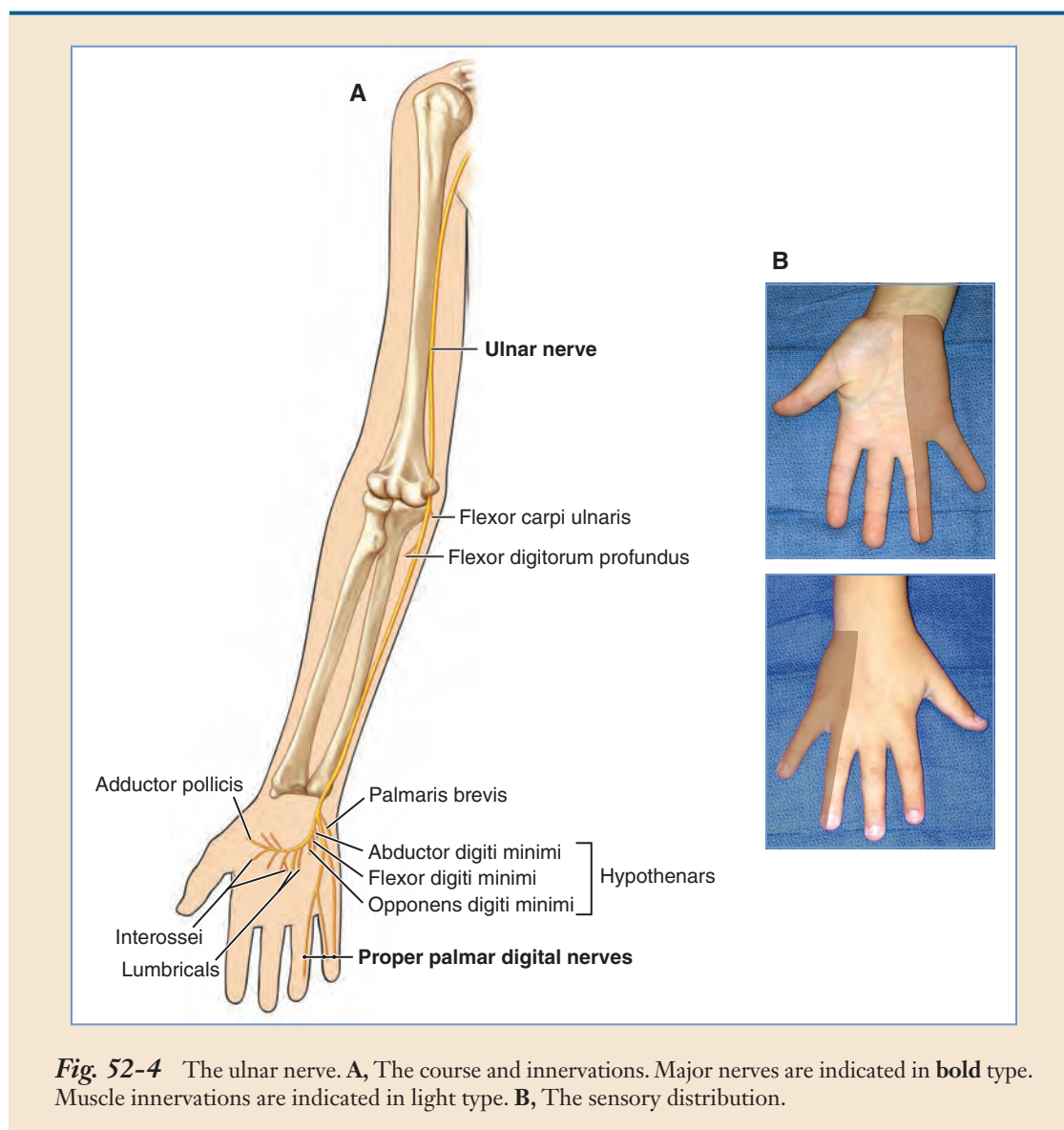


Fig. 52-3 The median nerve. **A**, The course and innervations. Major nerves are indicated in **bold** type. Muscle innervations are indicated in light type. **B**, The sensory distribution.

The median nerve then travels just beneath the palmaris longus tendon toward the palmar wrist. At this location it is quite superficial and vulnerable to injury. The nerve enters the hand through the carpal tunnel. At the level of the wrist, the median nerve motor fibers are located palmar and radial to the sensory fibers. The motor branch of the median nerve provides innervation to the thenar muscles and to the first two lumbrical muscles. The palmar cutaneous branch usually begins 6 cm proximal to the palm, travels radial to the palmaris longus tendon, and supplies sensation to the radial palm.

The ulnar nerve travels in the medial aspect of the arm (Fig. 52-4). It enters the forearm between the two heads of the flexor carpi ulnaris, having passed through the cubital tunnel on the medial aspect of the elbow. At this point, it is unprotected by any overlying musculature and is subject to trauma. The ulnar nerve runs between and innervates the flexor carpi ulnaris and the



two ulnar muscle bellies of the flexor digitorum profundus. Thus injury at this level will lead to motor deficits (the inability to flex the distal interphalangeal [DIP] joint of the small finger) and sensory deficits.

In the forearm, the ulnar nerve is topographically organized into three large and distinct bundles: the dorsal cutaneous branch, the superficial sensory branch, and the deep motor branch. Approximately 5 cm proximal to the pisiform bone, the ulnar nerve gives off the dorsal cutaneous branch, which supplies sensation to the ulnar side of the dorsal hand. The nerve enters the palm through the Guyon canal, where the superficial sensory branch splits into a common digital nerve sensory branch that gives off the common digital nerve to the fourth web space and the ulnar digital nerve to the little finger. The motor fibers (deep motor branch) of the ulnar nerve are located ulnar and dorsal to the sensory fibers and innervate all of the intrinsic muscles of the hand except those innervated by the median nerve.

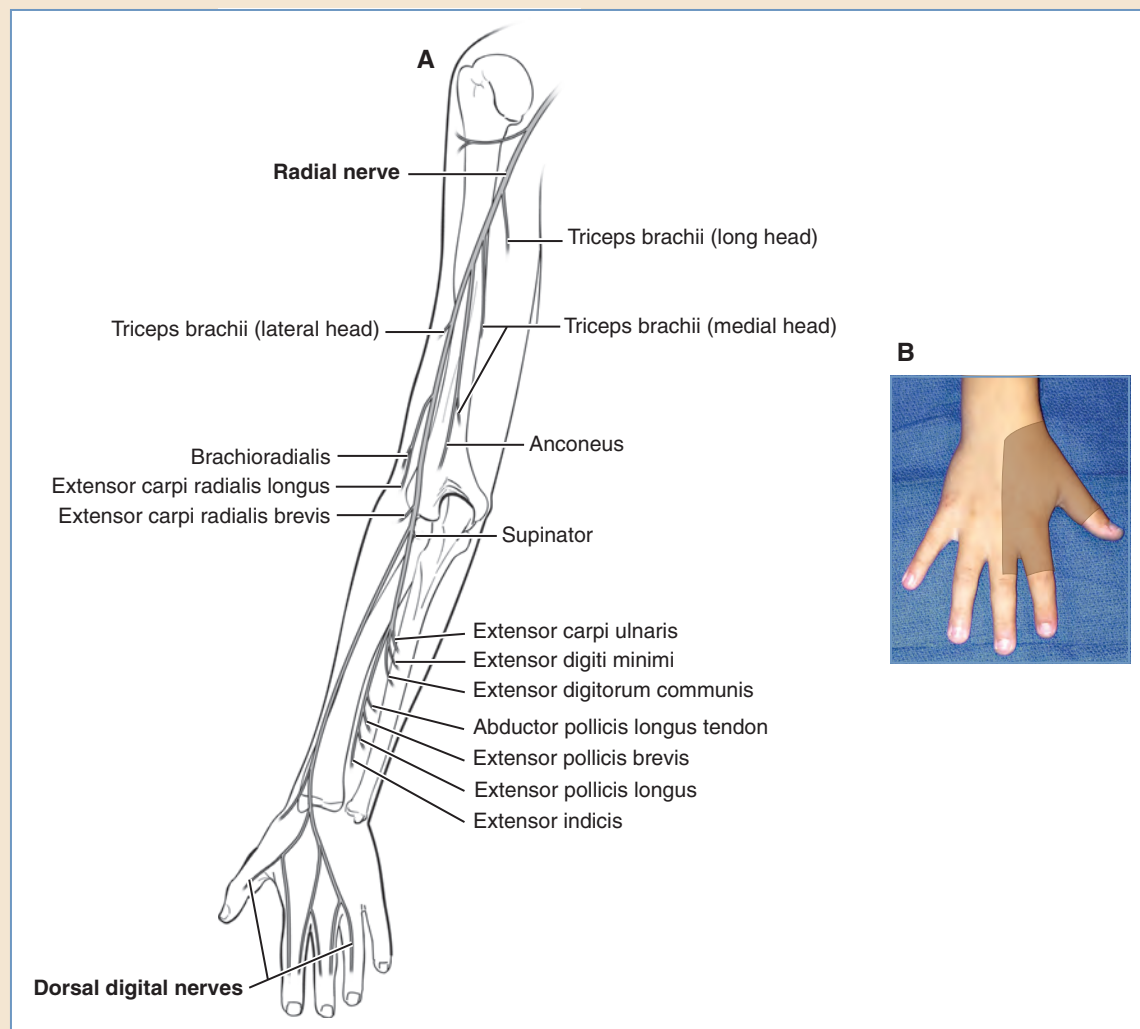


Fig. 52-5 The radial nerve. **A**, The course and innervations. Major nerves are indicated in **bold** type. Muscle innervations are indicated in light type. **B**, The sensory distribution.

The radial nerve in the arm travels from medial to posteroradial and then anteriorly between the biceps and brachialis muscles (Fig. 52-5). It travels on the shaft of the humerus in the spiral groove. The nerve courses anteromedially between the brachialis and brachioradialis over the lateral epicondyle. In the arm, the radial nerve innervates the triceps, the brachioradialis, and the extensor carpi radialis longus. The nerve continues anterolaterally into the forearm and divides into two major branches: the superficial and the deep (posterior interosseous nerve). The superficial radial nerve is composed mostly of sensory branches to the radial side of the hand. It often provides motor innervation to the extensor carpi radialis brevis. The posterior interosseous nerve runs through the two heads of the supinator and innervates the supinator, extensor carpi ulnaris, extensor digitorum communis, abductor pollicis longus, extensor pollicis brevis, extensor pollicis longus, extensor indicis proprius, and the extensor digiti minimi muscles.

The sensory innervation of the hand is supplied by all three nerves. In the fingers, the digital nerves lie palmar to the arteries on the radial and ulnar sides of the flexor tendon sheath. Therefore, when a penetrating injury leads to pulsatile bleeding, both the nerve and the deeper artery have probably been injured. At the level of the DIP joint, the nerves trifurcate into terminal branches to the tip of the fingers.

Muscles and Tendons

Flexor Muscles and Tendons

The muscles on the flexor aspect of the forearm can be divided into deep and superficial compartments (Figs. 52-6 and 52-7). The tendons insert onto the wrist or digits. The superficial compartment contains the palmaris longus, flexor carpi radialis, and flexor carpi ulnaris muscles. Proximally, the pronator teres is the deeper muscle belly. The flexor digitorum superficialis lies

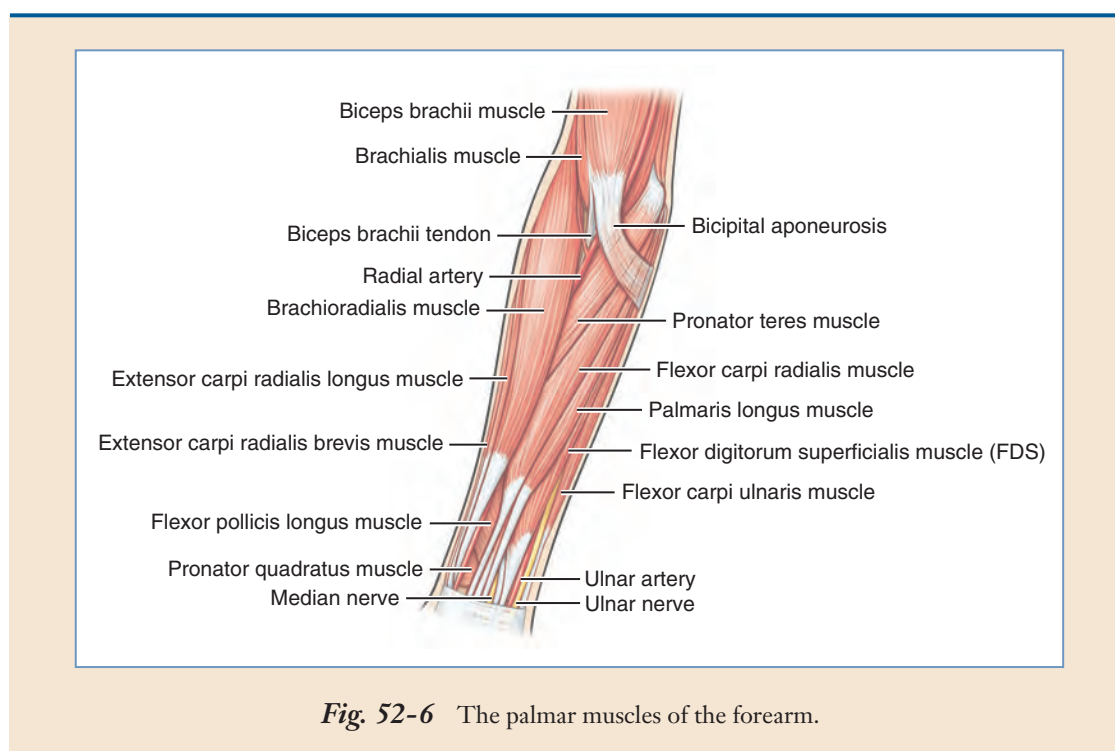


Fig. 52-6 The palmar muscles of the forearm.

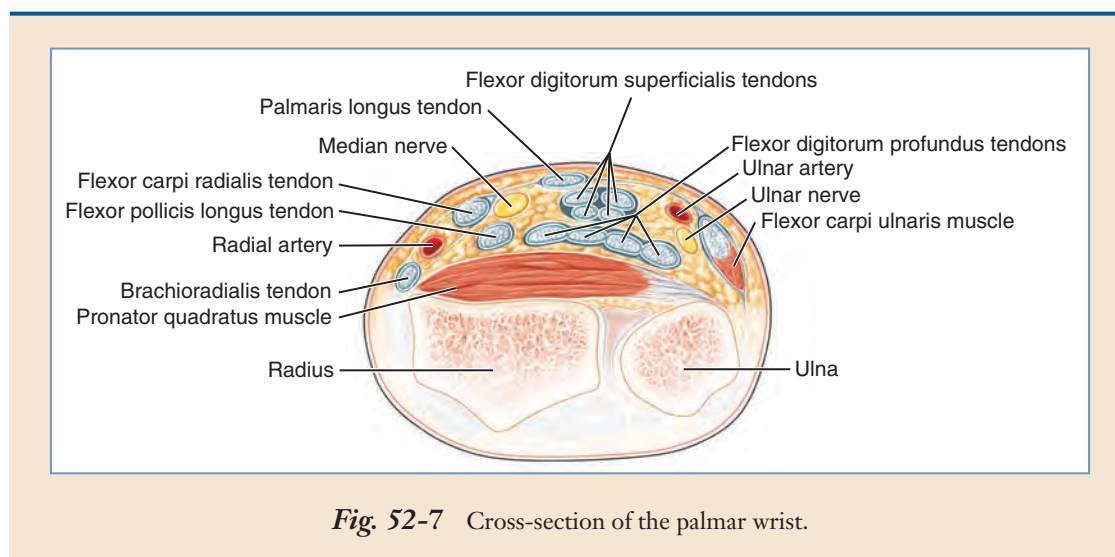


Fig. 52-7 Cross-section of the palmar wrist.

deep to this layer. The flexor digitorum superficialis arises from two heads and has four distal tendons that cross the wrist and flex the proximal interphalangeal (PIP) joints of the index, middle, ring, and little fingers. The tendons of the middle and ring fingers are more superficial than those of the index and little fingers at the level of the wrist. After the tendons form the Camper chiasm (through which the tendon of the flexor digitorum profundus travels), they insert into the middle phalanx of the fingers.

The deep muscle compartment in the forearm includes the flexor digitorum profundus and the flexor pollicis longus. Because these are in close contact with the bones of the forearm and the interosseous membrane, they are at the highest risk of having ischemic injury in compartment syndrome. The flexor digitorum profundus has four tendons, which cross the wrist and flex both the PIP and DIP joints of the respective fingers. Fascial connections are present between these tendons in the wrist and proximal to the wrist itself. The tendons of the flexor digitorum profundus insert into the base of the distal phalanx. In about a third of hands, the flexor digitorum superficialis to the little finger is absent. Usually, this deficit is unilateral. In the deep compartment of the distal forearm, the pronator quadratus spans the palmar aspect of the distal radius to the ulna. The flexor pollicis longus travels on the radial aspect of the carpal tunnel, then through the thenar eminence and into the fibroosseous canal to insert into the base of the distal phalanx of the thumb.

Extensor Muscles and Tendons

The extensor (dorsal) forearm contains the extensors of the wrist and the extrinsic extensors of the fingers (Figs. 52-8 and 52-9). The extensors arise from a common origin on the lateral epicondyle of the humerus. Arising deep and distally are the extensors and abductors of the thumb. At the level of the wrist, the extensor tendons travel through well-defined fascial compartments under the extensor retinaculum. There are six extensor compartments. The first compartment includes the abductor pollicis longus and extensor pollicis brevis. Although a particular pathology affects this compartment in adults, it is extremely rare in children. The second compartment includes the wrist extensors: the extensor carpi radialis longus and the extensor carpi radialis brevis. The third compartment contains the extensor pollicis longus, which extends the thumb and inserts on the thumb distal phalanx. The fourth dorsal compartment includes the finger extensors—the

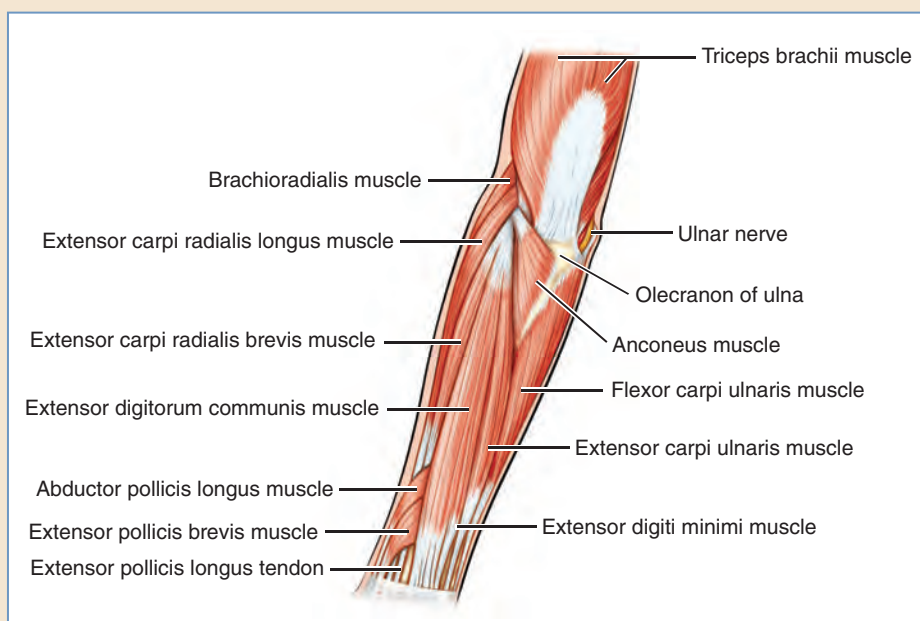


Fig. 52-8 The dorsal muscles of the forearm.

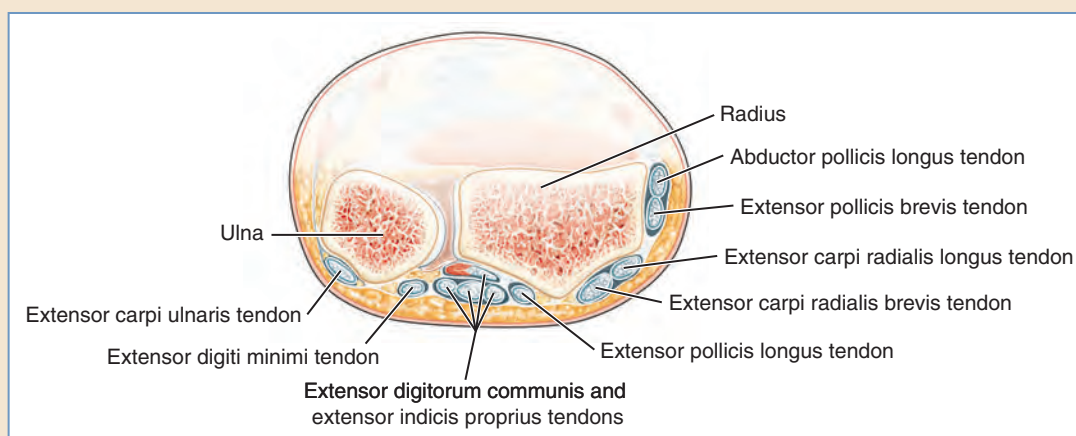


Fig. 52-9 Cross-section of the wrist at the distal forearm.

tendons of the extensor digitorum communis and the extensor indicis proprius. The fifth compartment includes the extensor digiti minimi, and the sixth includes the extensor carpi ulnaris.

Intrinsic Muscles

The intrinsic muscles of the hand are those that originate and insert within the hand. These include the lumbricals and the palmar and dorsal interossei. The four dorsal interossei are abductors of the fingers. The three palmar interosseous muscles are adductors. These muscles also flex

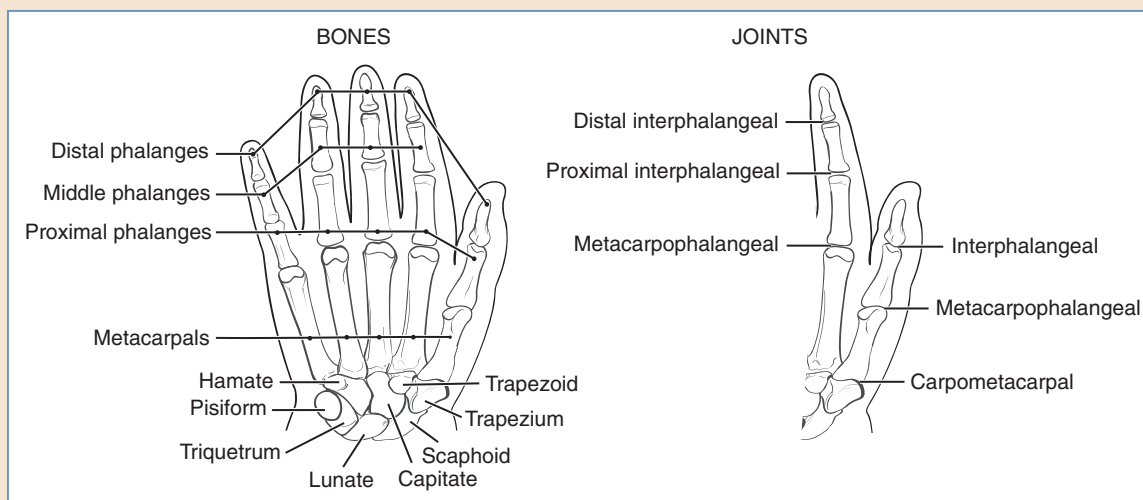


Fig. 52-10 The bones of the hand and wrist.

the metacarpophalangeal (MP) joints and extend the interphalangeal joints of the fingers. The muscles of the thenar and hypothenar eminences include the following:

- Abductor pollicis brevis
- Flexor pollicis brevis
- Opponens pollicis
- Abductor digiti minimi
- Flexor digiti minimi
- Opponens digiti minimi

The intrinsic muscles also include the adductor pollicis.

Bones and Joints

The bones of the hand and wrist include the radius and ulna, the carpal bones, the metacarpals, and the phalangeal bones (Fig. 52-10). There are eight carpal bones: the proximal carpal row consists of the scaphoid, lunate, and triquetrum; the distal carpal row includes the trapezium, trapezoid, capitate, and hamate; and the pisiform is a sesamoid bone within the tendon of the flexor carpi ulnaris.

The wrist joint comprises the distal radioulnar joint, radiocarpal joints, intercarpal joints, and the carpometacarpal joints. In each finger the proximal joint is the MP joint; the PIP and DIP joints are more distal.

PATIENT EVALUATION

As in all cases of trauma, the whole patient should be evaluated first, and the management of the injuries should be prioritized accordingly. A pediatric patient's general condition can rapidly deteriorate; therefore urgent problems or injuries should be addressed expeditiously. The primary goals of the initial evaluation in the emergency department are the following:

1. To determine what structures are injured
2. To determine whether the injury requires urgent or elective operative treatment
3. To provide the patient and parents with information on the injury severity, management goals, and expected prognosis
4. To assure the patient and parents that the injury will be cared for by an experienced, competent, and compassionate surgeon

We do not wear white coats during the consultation and find it very helpful to have the child sit in the parent's lap for reassurance. Beginning the evaluation slowly, gently, and in a playful manner is critical to secure the child's trust. Distraction techniques can be useful to make the child feel more at ease. In our institution, for example, we routinely show popular cartoon videos in each examination room during the evaluation. However, occasionally, a child is uncooperative to the extent that an evaluation cannot be performed. Intravenous sedation may be necessary in these situations.

A formal evaluation consists of a history, physical examination, and laboratory testing as necessary. The history should include specific information such as the mechanism of hand trauma, the time of injury, and the place of occurrence. A brief medical history is obtained to determine allergies and general conditions such as diabetes. A past surgical history is obtained, as well as any family history of medical illnesses, including anesthesia issues. Obtaining a social history helps in understanding the child's social support system and exposure to tobacco, alcohol, and drugs.

The physical examination should start with observation, followed by palpation, which can provoke apprehension and pain. Especially in children, this part of the examination is invaluable. We routinely first examine the unaffected extremity. This technique calms the patient significantly by helping the child understand what to expect. Next, before evaluating the injured extremity, the surgeon should reassure the child that the examination will stop if "it hurts too much."

A vascular assessment should be performed early in the examination, because vascular compromise will require urgent operative treatment. Vascularity can be evaluated by observing the color and capillary refill in the fingers. A temperature assessment is useful, especially a comparison between the injured and uninjured extremity. Pulses are palpated and pulp turgor assessed.

Sensation is sometimes more difficult to assess and requires the cooperation of the patient. This is not always possible, especially in small children. If possible, static and moving two-point sensation, paresthesias, and dysesthesias are documented. In very young children who cannot reliably communicate, alternative means can be used to assess nerve injury. In the presence of a nerve laceration, the skin overlying the distribution of the lacerated nerve feels dry, whereas uninjured regions are moist. A water immersion test can help to confirm a nerve injury (see Acute Nerve Injuries).

Motor function should be evaluated in the specific nervous distributions. Fractures and injuries to other soft tissues may affect the motor unit function performance in a nerve distribution, even though the nerves and neuromuscular structures are intact.

Swelling, deformity, and spontaneous use are noted. Palpation may help to localize an injury to bones and joints, especially in the wrist. Movement may be less sensitive and specific.

Myotendinous units can be examined in cooperative patients with no bony/joint injury. The flexors and extensors should be examined separately.

Flexor Tendon Examination

An examination of the flexor tendons of digits comprises the following four components:

1. Active range of motion
2. Observation at rest
3. A forearm squeeze test
4. The tenodesis effect

Of these tests, only the active range-of-motion test cannot be performed with the patient under general anesthesia.

Active Range-of-Motion Test

In an awake and compliant patient, the flexor digitorum profundus and flexor digitorum superficialis tendons can be examined separately. To examine the flexor digitorum superficialis, the fingers are examined individually by restraining the other four in extension and then the patient is asked to bend them in at the PIP joint. This maneuver neutralizes the flexor digitorum profundus, whose tendons are connected distally. The ability to fully flex at the PIP joint indicates a competent flexor digitorum superficialis tendon. The flexor digitorum profundus can then be examined by inhibiting flexion at the PIP joint of the finger and allowing flexion only at the DIP joint. The ability to flex at the DIP joint indicates an intact flexor digitorum profundus tendon. This examination is repeated, with gentle resistance applied. If attempted flexion is painful, a tendon injury should be suspected.

Observation at Rest

With the patient relaxed or under anesthesia, the posture of the digits can be observed. The normal cascade is of increasing flexion, beginning with the index finger and progressing to the small finger.

Forearm Squeeze Test

To test for a distal forearm injury, tendon continuity can be assessed by placing pressure on (squeezing) the proximal forearm. Intact myotendinous units will transmit the force and cause digit flexion. However, partial tendon lacerations can be misleading, because they may result in finger flexion.

Tenodesis Effect

To assess muscle-tendon continuity, the examiner evaluates the tenodesis effect (Fig. 52-11). Gentle, passive flexion and extension of the wrist cause passive digit extension and flexion, respectively, if the tendons are intact. Flexion should appear in the normal cascade.

Extensor Tendon Examination

An extensor tendon examination is composed of three basic components:

1. Active range of motion
2. Observation
3. The tenodesis effect

The most informative of the three examinations is an active range-of-motion evaluation. In a cooperative patient, each extensor muscle unit should be examined independently. The hand is

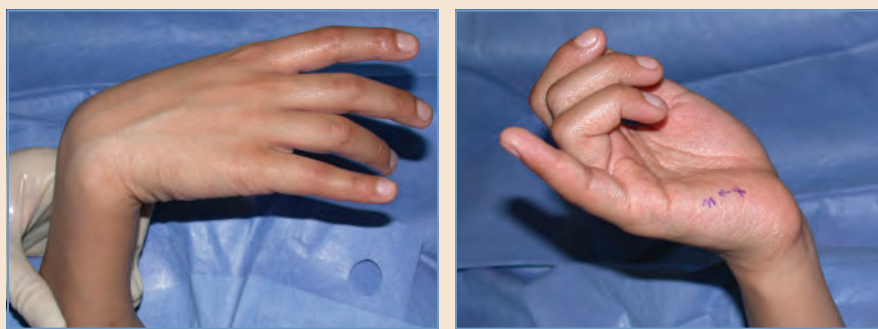


Fig. 52-11 The tenodesis effect in a patient with a zone II flexor digitorum superficialis and flexor digitorum profundus laceration. The small finger digital cascade is absent with the wrist passively extended.

allowed to rest comfortably, and the thumb and each finger is individually examined for MP, PIP, and DIP extension.

If it is determined that the patient is best cared for in the operating room, the injury should not be exposed in the emergency department. Except for superficial injuries, probing wounds in the emergency department is ill advised; without loupe magnification, tourniquet control, proper pain control, and adequate lighting it can prove harmful. Most decisions on the need for operative exploration can be based on a thorough history and physical examination.

FRACTURES IN THE PEDIATRIC HAND

Approximately 10% to 15% of all childhood injuries involve skeletal trauma.⁵ The growing skeleton often has a different mechanism of injury and a different capacity to heal and remodel. Therefore the treatment may vary with age and be different from that of an adult. Fractures and dislocations in the hand are less common in children than in adults. Fractures in children will heal faster than those in adults and will rarely develop stiffness after immobilization.

The growth plate (the epiphyseal plate and/or epiphysis) is the center of long-bone growth through the essential mechanisms for endochondral ossification.⁶ The anatomy of the epiphyseal plate is shown in Fig. 52-12. The primary function of the epiphysis (physis) is longitudinal and latitudinal growth of long bones. In the hand, the metacarpal and phalangeal bones are long bones. These bones have an epiphysis only on one end. The phalangeal physis is at the proximal end of the phalanx. The metacarpal physis is at the distal edge. Therefore most of the growth in the finger surrounds the MP joint. This is not the case in the thumb metacarpal, in which the epiphysis is located proximally. This difference may be helpful in distinguishing between a thumb and a finger in certain congenital deformities. Fig. 52-13 shows the time of appearance and fusion of the epiphyseal centers. An easy way to remember the times of epiphyseal center appearance is illustrated in Fig. 52-14. The epiphyses appear and close earlier in girls. A second epiphysis is sometimes present at the distal end of the first metacarpal or at the proximal end of the index metacarpal. This is referred to as a *pseudoepiphysis* and is sometimes confused with a fracture. These usually fuse early. Comparing radiographs of the affected side with those of the uninjured side can be helpful, because the epiphyses are irregular and individual differences occur. An atlas may not be readily available for reference.

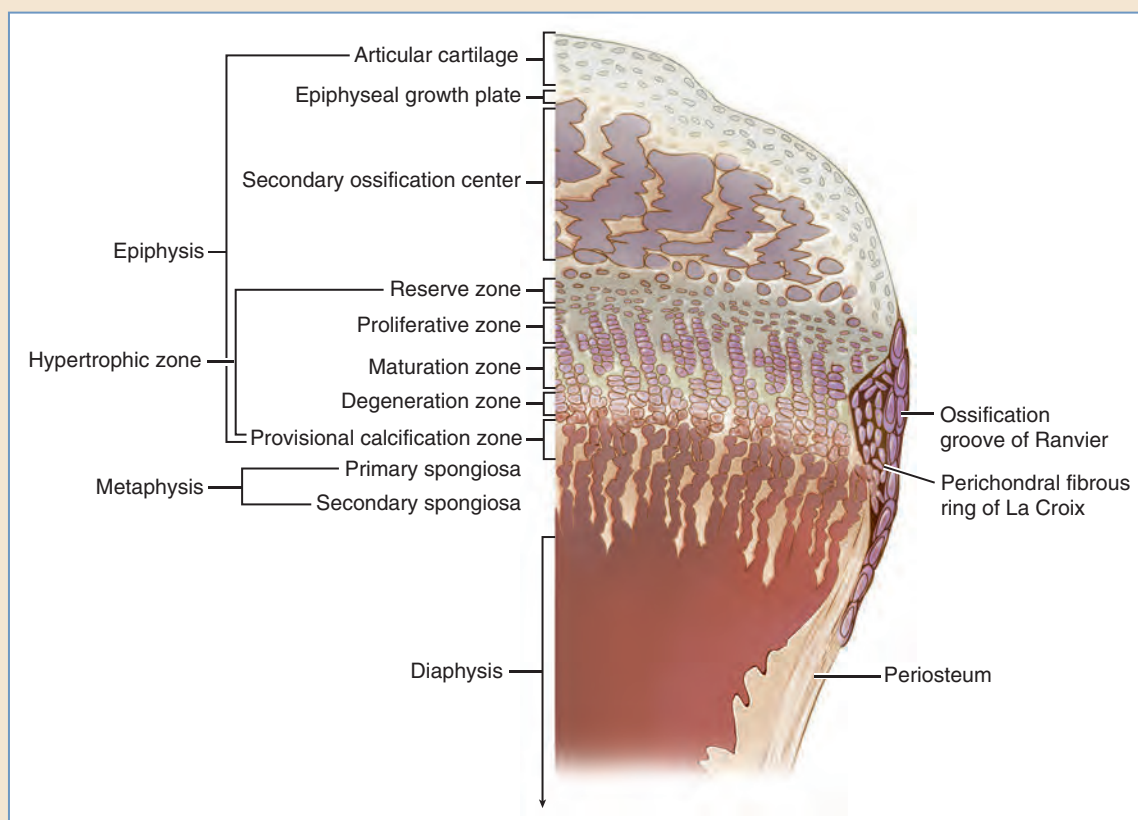


Fig. 52-12 Anatomy of an epiphysis.

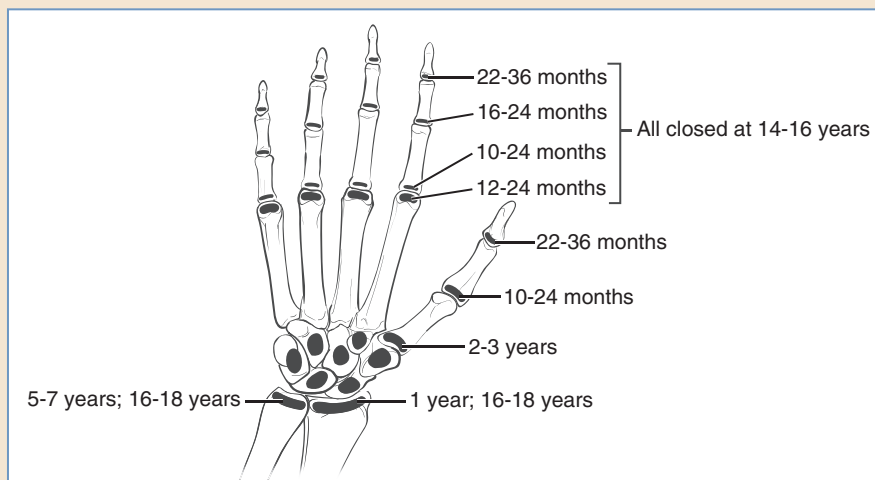


Fig. 52-13 The ages of epiphyseal center appearance and physeal fusion.

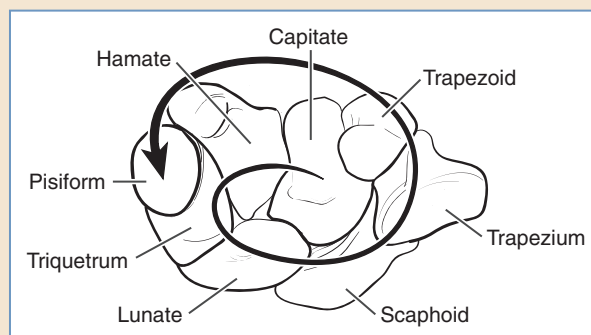


Fig. 52-14 Carpal bone appearance. The arrow denotes the usual sequence of carpal bone appearance: capitate, hamate, triquetrum, lunate, scaphoid, trapezium, trapezoid, and pisiform.

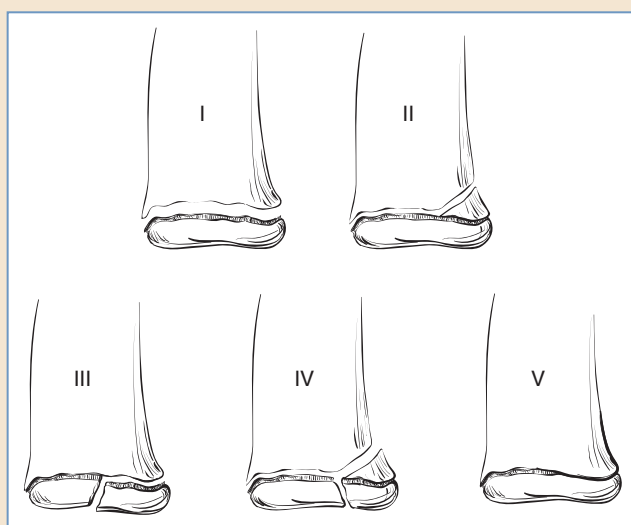


Fig. 52-15 The Salter-Harris classification system of epiphyseal fractures. (*I*, Separation of the epiphysis from the metaphysis; *II*, a fracture through the epiphyseal plate and the metaphysis; *III*, a fracture through the plate and the physis; *IV*, a fracture through the metaphysis, the epiphyseal plate, and the physis; *V*, a severe injury through the epiphyseal plate.)

Injuries involving the physis constitute about a third of all pediatric hand fractures.¹ These are areas of minor resistance; thus fractures can occur through them. Often, they are near insertions of ligaments or tendons. In the MP joint, the collateral ligament originates from the metacarpal epiphysis, and its cord portion inserts in the epiphysis of the proximal phalanx and the palmar plate. In the PIP joint, the collateral ligament inserts both into the epiphysis and into the metaphysis.

Evaluation

Radiographs should be obtained for all suspected fractures in the emergency department if the patient's general condition permits. Deformity may only be assessed with a physical examination, and patients should be examined clinically first. A rotational deformity especially may not be ap-

preciated on radiographs and is evaluated by observing the direction of the fingertips when the fingers are slightly flexed and comparing them with the uninjured side. Another technique is to examine the direction of the nail. This is often the critical factor in deciding treatment.

The evaluation and treatment of associated soft tissue injuries is critical to the healing of the fractures.

Classification

Fractures should be classified based on the following:

1. The anatomic position (that is, the bones involved and the area involved, such as the metaphysis)
2. Whether the fracture involves the articular surface, with or without dislocation
3. Whether the fracture is open or closed

The Salter-Harris classification⁷ is a very useful system for describing fractures involving the epiphysis (Fig. 52-15). This classification not only provides a means for communication but also aids in determining prognosis. Type I injuries include separation from the metaphysis. Salter-Harris type II is the most common injury and usually carries a benign prognosis.⁷ In this case, the force causing the fracture travels through the epiphyseal plate and the metaphysis. Type III fractures extend through the plate and the physis. Type IV fractures extend through the metaphysis, the epiphyseal plate, and the physis. Type V is a severe injury through the epiphyseal plate. The implication of this type of injury is a propensity to develop fusion of all or part of the growth plate. If the whole growth plate is affected, growth may be arrested. If only a part is affected, there is a risk of an angulatory deformity, because one area of the growth plate will grow more than the other. The parents should always be warned that growth might be affected. Although this is relatively rare, we do not have the means to accurately predict which fractures will undergo early fusion.

Table 52-1 Gustilo Open Fracture Classification

Type	Characteristics
I	A clean puncture wound of 1 cm or less No muscle contusion or soft tissue involvement No crush component
II	A laceration larger than 1 cm Without extensive soft tissue damage, flaps, or avulsions A minimal-to-moderate crush component
III	Extensive damage to soft tissues A high-velocity or severe crush injury Skin, muscle, or vascular injury
IIIA	Adequate soft tissue coverage
IIIB	Bone exposure and periosteal stripping
IIIC	An arterial injury requiring repair

Classically, open fractures have been described using the Gustilo classification⁸ (Table 52-1). Although this system was initially described for lower extremity injuries, the nomenclature has been adopted for upper extremity open injuries as well. Type I injuries are fractures with a wound of 1 cm or less. Type II fractures are open fractures with a wound larger than 1 cm but without extensive soft tissue damage. Type III fractures are associated with extensive soft tissue damage and have three categories: type IIIA has adequate soft tissue coverage, IIIB is associated with bone exposure and periosteal stripping, and IIIC has an arterial injury requiring repair.

Treatment

As in all hand fractures, treatment consists of reduction and fixation. Most fractures in the hand and wrist can be reduced in the emergency department, with the patient under a digital or wrist block. A splint or cast is applied. Minor degrees of angulation may correct with remodeling and should require no further treatment. This is especially true for displacement in the direction of joint motion. The younger the child, the greater the remodeling potential. Correction of rotational fractures is less reliable. Scissoring of the finger or abduction in a border digit after reduction, necessitates open reduction. Operative management is probably required. Epiphyseal injuries that are not easily manipulated should probably be treated in an open manner. In general, multiple attempts at reduction can cause permanent epiphyseal damage and are discouraged. After about a week, the fracture has begun to heal, and more damage can be done by trying to manipulate these fractures in a closed fashion. Immobilization on the whole is more difficult in children than in adults, particularly in small children, in whom a long cast may be required for a finger fracture. Open reduction is indicated when closed reduction cannot be achieved. Reductions that can be obtained but not maintained are considered unstable and require some manner of fixation.

The most common fractures are phalangeal—more often occurring distally. Most distal phalangeal fractures result from a crush injury to the fingertip and include soft tissue injury. The nail bed should be repaired with fine absorbable suture. Rarely, the nail bed becomes interposed in an epiphyseal fracture (Seymour fracture), necessitating an open reduction (Fig. 52-16). Seymour fractures that go unrecognized can result in malunion or nonunion, osteomyelitis, and permanent nail deformity. This injury should be considered with any apex dorsal fracture through the distal phalanx physis.

Phalangeal shaft fractures can be transverse, oblique, spiral, or comminuted and are treated according to the deformity and stability after closed reduction. Salter-Harris type II fractures of the proximal phalanx are common, especially in the little finger.⁷ These can usually be reduced in a closed fashion using a digital block. Placing a pen between the ring and little fingers may be helpful for reducing the fracture by stabilizing the proximal fragment (Fig. 52-17). The distal finger is then adducted toward the ring finger, bringing the distal fragment back into place (see Fig. 52-17). Metacarpal fractures in children usually do not require operative treatment. Metacarpal neck fractures are fairly common but rarely require surgery.

Fractures of the carpal bones are relatively rare, although scaphoid fractures occur at any age. Because many of the carpal bones are only partially ossified, these fractures often are difficult to diagnose and are classified as wrist sprains. In children, they are usually treated conservatively but with a longer immobilization period (6 weeks). As in adults, fractures of the scaphoid are probably the most common; the diagnosis is made from a history of a fall, often on an outstretched hand, and an examination that elicits tenderness in the area of the snuffbox. As in adults, such fractures in children, even if only suspected, should be treated with a thumb spica splint or cast, followed by repeat radiographs in 2 weeks. The incidence of nonunion is lower in children because of increased stability and their superior biologic factors.

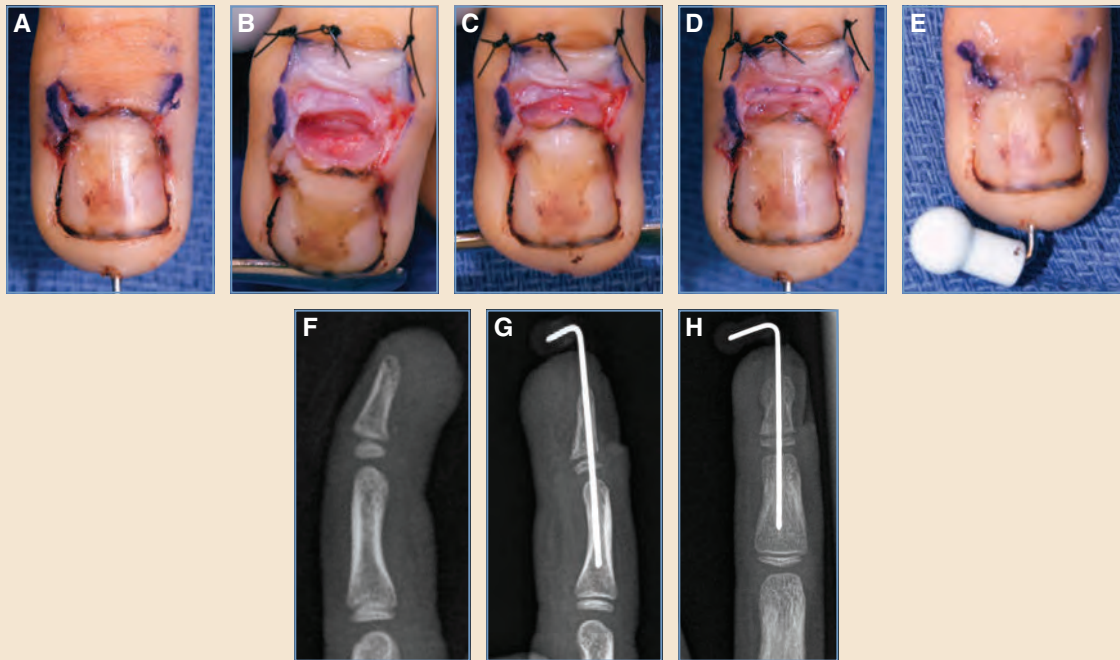


Fig. 52-16 Correction of a Seymour fracture. **A**, Incisions to reflect back the eponychial fold. **B**, Germinal matrix caught inside the fracture. **C**, Germinal matrix delivered out of the fracture gap. **D**, Germinal matrix repaired to the terminal tendon. **E**, Eponychial fold incisions repaired with cyanoacrylate glue. **F**, A preoperative radiograph shows an apex dorsal angular deformity through the physis. **G** and **H**, Postoperative radiographs show the pins in place.

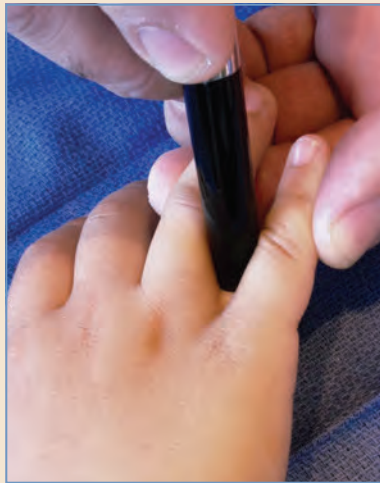


Fig. 52-17 Closed reduction of the proximal phalanx of the little finger using a pin to stabilize the proximal fragment while the distal part of the finger is adducted toward the ring finger. With this adduction pressure, the fracture will realign.

The most commonly fractured bone in children is the radius; 82% of these involve the distal third of the bone.⁹ A general classification of these fractures includes the following types: physeal, torus, greenstick, and complete. A reasonable closed reduction is usually obtained, and growth disturbances are rare, even with physeal involvement. Some degree of angulation can be corrected with growth and remodeling, especially when the fracture line lies close to the growth plate. With more proximal involvement of both bones, the reduction can be more difficult and require an open method.

DISLOCATIONS

Dislocations can be associated with fractures, or they may be purely soft tissue injuries. They are classified as simple (reducible) or complex (irreducible, usually as a result of soft tissue interposition). Similar to fractures, they may be open or closed. Dislocations in a child's hand have to be distinguished from fractures that include the epiphyseal plate. This distinction can only be made with radiographs.

The MP and interphalangeal joints tend to dislocate dorsally (that is, the distal bone dislocates dorsally). Most simple dislocations in the hand can be reduced in the emergency department. In the thumb, an often-irreducible fracture/dislocation is the Bennett fracture equivalent. This is a fracture dislocation of the base of the thumb. Compared with adults, children often have a larger fragment of bone, and the epiphysis is not always clearly involved. However, this fracture behaves like an adult Bennett fracture in that the distal/dorsal fragment is pulled proximally and radially by the tendon of the abductor pollicis longus, and the proximal/palmar fragment remains attached to the palmar oblique ligament of the joint. These fractures are surprisingly difficult to reduce and must be held in the reduced position.

Conservative treatment consists of 3 to 4 weeks of immobilization. The younger the child, the less time necessary for immobilization. The hand should be in the intrinsic-plus position within the cast, with the MP joints at 70 or 80 degrees of flexion and the interphalangeal joints in full extension. Operative treatment for fracture reduction and stabilization is usually minimal; the surgeon should disturb the growth plates as little as possible. Plates are therefore rarely used; smooth K-wires are the mainstay of treatment.

TENDON INJURIES

Flexor Tendon Injuries

The treatment and repair of flexor tendons in children differs from that in adults in these ways^{10,11}:

- Especially in small children, initial immobilization and later early mobilization according to adult protocol may be difficult, if not impossible.
- Possibly for this reason, but also because of the more rapid fibroblast proliferation in children, contractures after repair are often more pronounced and more difficult to overcome.
- Because of the relatively small size of the different structures, the technical aspects of the repairs are more challenging.

The zone of injury is significant both for repair and for functional outcome. The classification system, a modification of the Verdan system,¹² is a useful tool for relaying information regarding an injury (Fig. 52-18). In this system, flexor zone I includes the flexor digitorum profundus alone and its insertion into the distal phalanx. Zone II includes the area beneath the fibroosseous sheath, which includes both the tendon of the flexor digitorum profundus and the flexor digitorum superficialis. This area is called *no-man's land* because of the poor postoperative functional

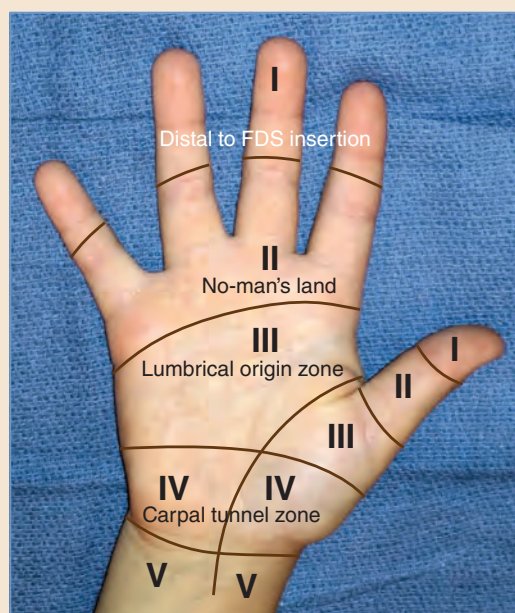


Fig. 52-18 Verdan zones of flexor tendon injury. (See the text for descriptions of all zones.)

results after tendon repair in this area. Zone III is the area between the carpal tunnel and the entrance to the fibroosseous sheath. This area contains the origins of the lumbrical muscles. Zone IV is the area of the carpal tunnel. Zone V is the area proximal to the carpal tunnel and includes the myotendinous junctures.

In the thumb, zone I is similar to that in the fingers and includes the edge of the retinaculum to the insertion of the flexor pollicis longus. Zone II includes the area under the flexor retinaculum of the thumb from the neck of the metacarpal to the neck of the proximal phalanx. Zone III includes the tendon proximally in the thenar eminence. These zones relate to the level of tendon injury and not to skin laceration.

The skin injury does not always correlate with the level of tendon injury. The level of tendon injury depends on the degree of finger flexion at the time of injury. The injury is repaired in the operating room, with exploration of the digital nerves. Better results are achieved if the repair is done within 10 days to 2 weeks after the injury. The skin incisions (for example, a zigzag Bruner incision,¹³ a Z-plasty incision, or midlateral incisions in the fingers), as in all palmar injuries, should allow healing without longitudinal contraction.

Flexor tendons should be repaired with a core suture so that gliding is not hindered. However, the type of suture used is controversial and depends on the surgeon's preference. Most surgeons supplement the core sutures with a running epitendinous suture. This has been shown to increase the strength of the repair in addition to "tidying" the repair edges. Rarely, an injury is infected or crushed to a degree that a delayed repair is deemed necessary.

After the tendon repair, protected immobilization is the recommended rehabilitation principle. In young children, a long-arm "clamdigger" cast is placed, with the elbow in 95 degrees of flexion, the wrist in 0 to 20 degrees of extension, the MPs in 60 to 70 degrees of flexion, and the interphalangeal joints in full extension. A short-arm clamdigger cast is applied for older children. We routinely place thick gauze padding from the MP joints to the fingertips and remove the

padding once the cast has hardened. This maneuver provides space for protected active range of motion for the digits within the confines of the cast.¹⁴⁻¹⁷

The extremity is immobilized for 4 weeks. In children younger than 3 years of age, no further immobilization is required. In patients older than 3 years of age, a dorsal blocking splint is worn during the daytime (to prevent accidental forceful digit extension) for an additional 3 weeks. Thereafter, our hand therapists teach the parents flexion and extension play therapy exercises so that they can begin a home therapy program. If by postoperative week 8 the child is unable to fully extend the digit, a formal hand therapy program is initiated with nighttime extension splinting. In adults, when adhesions limit digital function and an ample course of hand therapy has reached maximal usefulness, a flexor tenolysis is considered. In our experience, tenolysis in children is technically most challenging, and only minimal postoperative improvement is expected. Therefore we do not routinely perform flexor tenolysis for children younger than 10 years of age.

Extensor Tendon Injuries

The anatomy of the extensor mechanism—especially in the finger—is complex and very difficult to reproduce correctly. As in the diagnosis and treatment of flexor tendon injuries, extensor tendon injuries are classified into Verdan zones¹² (Fig. 52-19). Zone I is the area over the DIP joint, zone II is the area over the dorsum of the middle phalanx, zone III is over the PIP joint, zone IV is over the proximal phalanx, zone V is over the metacarpophalangeal joint, zone VI is over the metacarpal, zone VII is over the carpals, zone VIII is over the proximal wrist, zone T-I is at the interphalangeal joint, zone T-II is over the proximal phalanx, zone T-III is at the metacarpophalangeal joint, zone T-IV is at the metacarpal, and zone T-V is over the carpometacarpal joint. The areas of injury proximal to the finger are more easily reconstructed and can have very good functional results, especially in children.



Fig. 52-19 Verdan zones of extensor tendon injury. (See the text for descriptions of all zones.)

Zone I injuries cause an inability to extend the distal phalanx, resulting in a mallet finger. The most common mechanism of injury is jamming of a fingertip. These often occur in sporting activities like football, volleyball, and basketball. The injury can result in a tendon rupture or avulsion fracture of the extensor tendon insertion into the distal phalanx. In children, bony mallets are more common than nonbony mallets and often involve the epiphyseal plate, because this is the area of minimal resistance. In a typical closed mallet finger deformity, the terminal extensor tendon is thin and not able to accept suture. Furthermore, the ruptured ends do not usually retract far. Therefore most of these injuries can be treated by closed reduction and 6 to 8 weeks of continuous splinting with extension of the DIP joint. Avulsion injuries with a dorsal bony fragment that is a third of the articular surface, or if articular subluxation and instability are present, operative intervention is indicated to restore the articular architecture. Following the 6- to 8-week immobilization period, gentle protected digital range-of-motion exercises are initiated, and the DIP joint is splinted at night for another 2 to 4 weeks. However, if after 6 to 8 weeks of immobilization an unacceptable extension lag persists, up to 6 more weeks of uninterrupted splinting may be required.

Zone II injuries are similar to those in zone I and result in a flexion deformity of the DIP joint. The extensor mechanism is more robust in this region compared with that of zone I, and clean lacerations can usually be repaired with 5-0 nylon horizontal mattress sutures. Segmental losses are more difficult to repair and require reconstruction. Postoperatively, the DIP joint is immobilized by K-wire or splinting for 4 to 6 weeks.

Zone III is located over the PIP joint; it is a complex region where the components of the extensor mechanism merge, including the central slip and the lateral bands. Injuries in this area can be caused by laceration, avulsion of the middle phalanx dorsal articular surface, and PIP fracture dislocations. Injuries are often misdiagnosed, and if left untreated, result in a deformity in which the head of the proximal phalanx protrudes through the extensor hood, and the lateral bands progressively palmarly displace. This progressive deformity results in a boutonnière (buttonhole) deformity with a flexed PIP and a hyperextended DIP posture (Fig. 52-20).

With acute open injuries, the PIP joint capsule requires irrigation and separate closure. The central slip is repaired or reattached to the middle phalanx, and the lateral bands are medialized. The digit is immobilized for 1 week by placing the MP joint in 30 degrees of flexion, the PIP joint in full extension, and the DIP joint is left free to allow full motion. After 1 week, only the PIP joint is immobilized in full extension for 5 weeks.

Closed ruptures of the central slip are best treated by PIP extension splinting for 4 to 6 weeks. With the DIP joint free, the patient can pull the lateral bands up while flexing the DIP joint, improving the clinical result.

Injuries in zones IV and V are treated similarly to those in zone III. The tendon is repaired with 4-0 or 5-0 nylon sutures with a horizontal mattress or figure-of-eight technique. For best



Fig. 52-20 A chronic boutonnière deformity from an untreated central slip injury.

postoperative results, lacerations of the index and small finger extensor indicis proprius require repair despite the presence of a competent extensor indicis proprius and extensor digiti quinti tendons. Postoperatively, the repair is protected by immobilization for 3 to 4 weeks. Although in adults there has been a trend toward early active motion, this is usually difficult to accomplish in children; it is also unnecessary.

Injuries in zones VI and VII are treated similarly. However, zone VII injuries lie within the fibroosseous extensor compartments and can be challenging to manage. Tendons are repaired with 4-0 nylon or polyester suture using a figure-of-eight or modified Kessler technique.¹⁸⁻²⁰

Thereafter, the repair is passively tested to ensure there is no impingement through the canal. Significant excursion obstruction requires excision of part of the extensor retinaculum, or, if necessary, the tendon can be transposed outside of the canal. Postoperatively, the repair is protected by immobilization of the wrist for 4 weeks in 30 degrees of extension and the MP joints in 45 degrees of flexion, followed by protected passive range-of-motion exercise.

Zone VIII injuries can occur at the myotendinous junction or involve the muscle bellies. Similar to the other zones, nonabsorbable suture with a modified Kessler figure-of-eight technique is used for repair. To add strength to the repair, the muscle epimysium is included in the suturing. Postoperatively, the wrist is immobilized in 30 degrees of extension for 4 to 6 weeks, followed by passive range-of-motion exercises.

FINGERTIP INJURIES

Fingertip traumas are among the most common hand injuries, especially in toddlers.^{4,21} Young children's probing fingers are often injured in house and car doors, bicycle chains, and home exercycles.^{22,23} Recognition and proper management are essential for maintaining hand function and preventing disability. Surgical management should attempt to preserve length and restore sensibility.

The fingertip is a specialized end organ that involves all structures distal to the insertion of both the flexor digitorum profundus and the terminal extensor tendon (Fig. 52-21). Tissues include the nail plate, nail bed, distal phalanx, subcutaneous fat, nerves, vessels, lymphatics, and skin. The germinal matrix gives rise to 90% of the nail plate, which is an epidermal structure consisting of keratinized squamous cells.²⁴ The nail plate provides improved pulp sensation and stabilization and serves a protective function. The nail plate and distal phalanx form a protective

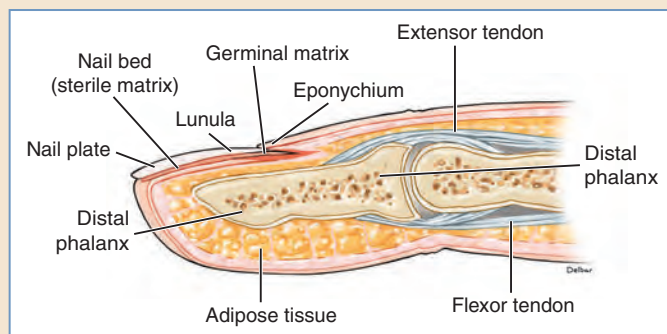


Fig. 52-21 Anatomy of the fingertip.

sandwich for the nail bed. The nail bed adheres to the nail plate by longitudinal interdigitating ridges produced by the sterile matrix. The nail grows at the rate of 0.1 mm per day and requires nearly 6 to 9 months to achieve full growth.²⁴ After a traumatic injury, nail growth is halted for 3 weeks, followed by a 7-week period of increased nail formation.²⁵ Most injured nails achieve their final aesthetic appearance after approximately 1 year.

The fingertip is highly vascularized, densely innervated, and adapted for pinch, grasp, and manipulative functions. The specialized palmar glabrous skin contains a series of ridges (fingerprints) that provide a nonslip surface that is stably attached to the distal phalanx through dense, vertically oriented septa.²⁶ The finger pulp contains fibroadipose tissue that serves a padding function and is compartmentalized by the vertically oriented septa. Bilateral digital arteries and nerves traverse the soft tissues and arborize into the pulp. The digital nerves run palmar to the digital arteries and trifurcate near the level of the DIP joint, sending branches to the paronychium, fingertip, and palmar pulp.²⁷

Hematoma and Nail Bed Injuries

Trauma to the nail bed includes lacerations, crush injuries, avulsions, and hematomas. Injuries that involve the germinal matrix may result in nail absence, whereas injuries to the sterile matrix can cause nail plate deformity.

Hematoma

An injury to the fingertip can result in a nail bed laceration and a collection of blood beneath the nail plate, with the development of a painful subungual hematoma that can potentially compromise the survival of the nail bed. If the hematoma size is more than 25% of the nail plate and a distal phalangeal fracture is evident, removal of the nail plate, followed by inspection, is recommended.²⁸ However, some authors advocate that the decision to remove the nail plate should not be based on the percentage of hematoma, but on whether the nail edges are intact.²⁹ If the nail edges are intact, simple evacuation of the hematoma is sufficient. However, if blood has undermined the nail plate and the edges are not adherent or the nail is broken, the nail plate should be removed.³⁰

To evacuate a hematoma, the digit is prepared and draped in a standard surgical fashion and anesthetized by a finger block with 1% plain lidocaine. A battery-powered disposable ophthalmic cautery device, available in most emergency departments, can be used to burn a hole in the nail plate; alternatively, a heated paper clip can be used (Fig. 52-22). Care is required to prevent injury to the underlying nail bed, and the hole should be large enough to allow continued drainage of the hematoma.

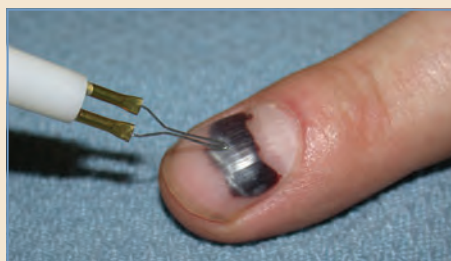


Fig. 52-22 Evacuation of a subungual hematoma with a handheld disposable ophthalmic cautery device.

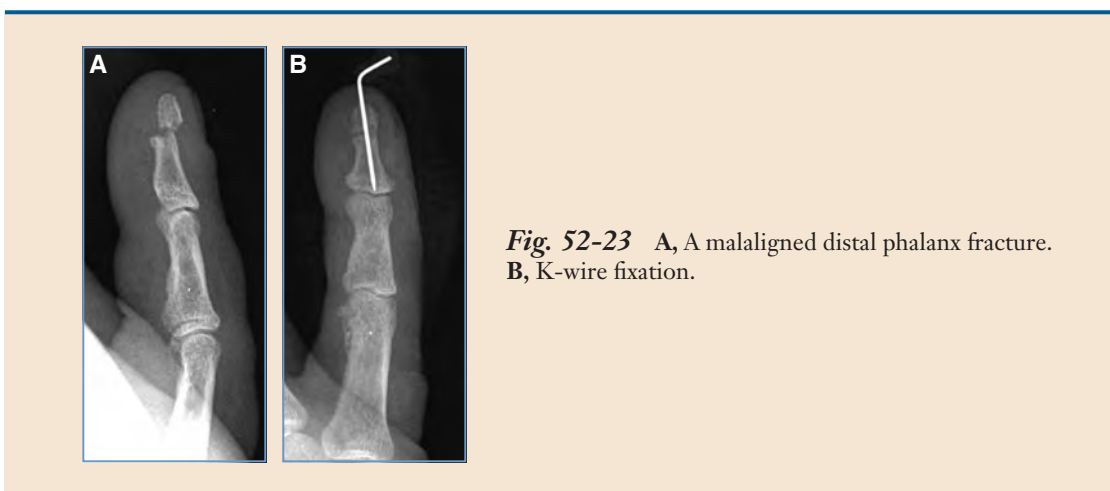


Fig. 52-23 A, A malaligned distal phalanx fracture. B, K-wire fixation.

Nail Bed Lacerations

Nail plate deformities can persist if trauma to the nail bed is not repaired anatomically. Radiographs of the distal phalanx and the nail bed are obtained to assess the anatomy. Most injuries can be comfortably addressed in the emergency department. After the patient is prepared and anesthetized with a digital block, a proximal phalanx finger tourniquet is applied for hemostasis.³¹ The nail plate can be gently separated from the nail bed using a Freer elevator or Iris scissors, with the tips pointing up, and proceeding from distal to proximal. Under loupe magnification, the nail bed is inspected and lacerations repaired with 6-0 or 7-0 absorbable plain gut sutures. The nail plate is cleaned with povidone iodine (Betadine) solution and irrigated with sterile saline solution. The fibrous undersurface attachments are scraped off. The nail plate is then replaced to stent open the eponychial fold, to provide protection for the nail bed, and to serve as a splint for distal phalanx fractures. If the native nail plate is not available, a nail-shaped stent can be fashioned from the aluminum suture package. The nail plate or stent should be secured with 5-0 chromic sutures, though this is frequently not required, and covered with nonadherent dressings.

Associated distal phalanx fractures are common findings with nail bed lacerations. Small tuft fractures do not require fixation, and the replacement of the nail plate serves as an acceptable splint. Larger, displaced fragments may be fixed with 0.028-inch K-wire (Fig. 52-23). Open fractures should be irrigated with sterile saline solution, reduced as best as possible, immobilized with a splint or K-wires, and treated with antibiotics. To best immobilize the distal phalanx, a splint is fashioned that includes the DIP joint; this is worn for 2 to 3 weeks or until the pain subsides.

Fingertip Amputations

The goals for the treatment of fingertip amputations are to preserve functional length, to restore adequate sensation, and to regain pain-free range of motion. Most surgeons agreed that preserving thumb length is critical; however, they agree less about the value of maintaining length in the other digits. Although numerous techniques have been described for fingertip reconstruction, ranging from soft tissue closure by secondary intention to microsurgical replantation, each injury should be considered individually and treatment plans made on a case-by-case basis. Several schemes have been proposed to help categorize fingertip injuries.^{8,32-34} The Allen classification⁹

of fingertip injuries is a simple and easy-to-apply system that categorizes amputations by the level of amputation (Fig. 52-24). Table 52-2 offers treatment options for each amputation level. To help better describe injuries, the Allen classification can be further divided by the planes of angulation (transverse, dorsal oblique angulation, and palmar oblique angulation).

Allen type I injuries involve only the skin and pulp of the distal digit. Children have remarkable healing potential, and these amputations can be appropriately and conservatively managed with excellent return of sensibility and cosmesis. Wound care consists of daily dressing changes with nonadherent dressings or petrolatum gauze. However, parents must be cautioned that wound healing may take 4 to 8 weeks. Alternatively, these wounds can be skin grafted for accelerated healing. Full-thickness glabrous skin hypothenar grafts are preferred over split-thickness grafts, because the cosmetic result is superior and the return of sensibility is reportedly better.

Allen type II injuries involve the pulp and the nail bed. Similar to type I injuries, most superficial type II injuries can be conservatively treated.

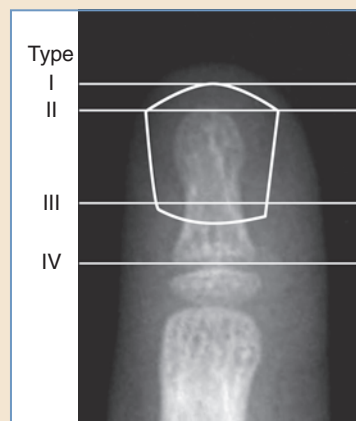


Fig. 52-24 Allen zones of fingertip injury. (See the text and Table 52-2 for descriptions of and treatment options for each zone.)

Table 52-2 Treatment Options by Amputation Level

Allen Type	Treatment Options
I	Conservative wound management Skin graft and/or allograft
II	Conservative wound management Skin graft and/or allograft Flap coverage Composite graft
III	Flap coverage Composite graft Closure of amputation
IV	Replantation Closure of amputation

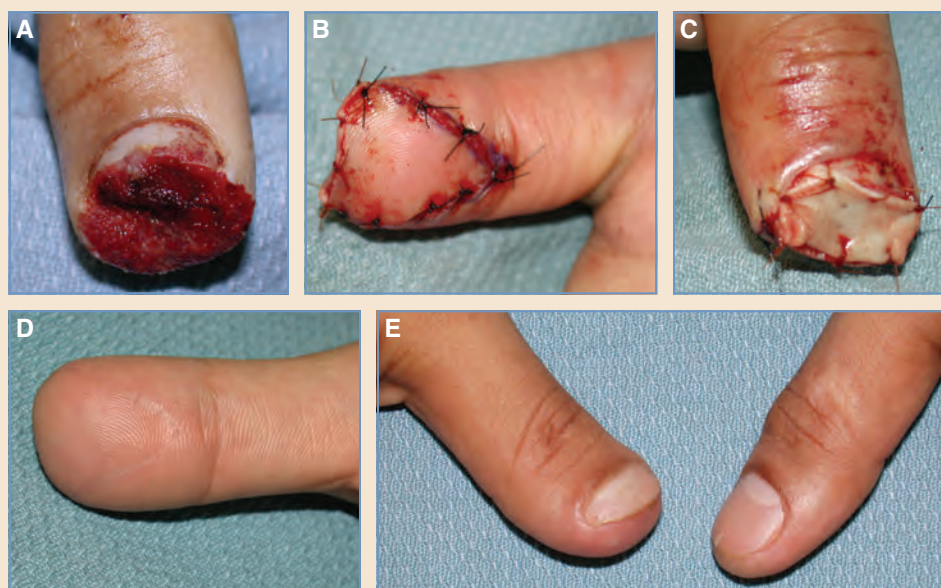


Fig. 52-25 A, A thumb distal tip amputation from a meat slicer. B, A palmar V-Y advancement flap. C, Flap perfusion appeared compromised by excessive tension; therefore the tension was relieved by allograft interposition. D and E, The 3-month postoperative result.

Allen type III injuries involve the distal phalanx, and type IV injuries are amputations proximal to the lunula. Deeper type II to IV injuries can be treated with skin grafts, local flaps, composite grafts, or replantation. If the amputation is sharp and not crushed, the soft tissue can be cleaned, defatted to the dermis, and attached as a composite graft. Composite grafts have the best chance for success if they do not exceed 8 mm in length and are used in children younger than 3 years of age. The graft should be sutured with 4-0 or 5-0 plain gut sutures, secured with a bolster dressing, and immobilized. Even if a composite graft does not fully survive, it will serve as a biologic dressing while the deep tissues granulate. Alternatively, a digital tip amputation stump can be covered with a local advancement flap, cross-finger flap, thenar flap, kite flap,³⁵ neurovascular advancement flap,^{36,37} or neurovascular island flap,³⁸ particularly for exposed bone. The most commonly used local advancement flaps are the palmar (Atasoy) V-Y^{39,40} flaps. Lateral (Kutler) V-Y flaps⁴¹ have been described; however, we have been disappointed with the results in children and no longer use them. These flaps provide limited advancement; if they are placed under excessive tension, flap necrosis may result. We recommend advancement of the flaps and inset with sutures. The tourniquet is then deflated, and the digit is irrigated with warm saline solution. If a flap does not develop acceptable perfusion, then a small piece of full-thickness skin graft or allograft is used to relieve flap tension (Fig. 52-25).

AMPUTATION AND REVASCULARIZATION

Revascularized digits and amputations proximal to the distal phalanx are considered surgical emergencies and require prompt attention. It is general consensus that amputation of any body part in a child is an indication for replantation if the amputated segment is suitable for replanta-

Box 52-1 Goals of Replantation and Revascularization

Restoration of hand function and joint mobility
Recovery of sensation
Minimization of pain
Optimization of final cosmesis

tion and the patient has no concomitant life-threatening injuries.^{42,43} The goals of replantation and revascularization are listed in Box 52-1.

Absolute contraindications to replantation include concomitant life-threatening injuries and underlying medical conditions that preclude undergoing a long operation. Relative contraindications for pediatric replantation include severe crush injuries, multilevel amputations, the patient's mental instability, and prolonged ischemia time. Because of cellular death and reperfusion injury, it is generally recommended that replantation not be performed when the warm ischemia time is longer than 6 hours for amputations proximal to the carpus and longer than 12 hours for the digits.⁴⁴ However, preservation of the amputated part at 4° C will provide more time to perform the replantation safely (12 hours for amputations proximal to the carpus and 24 hours for digits). This rule is particularly applicable for digits that do not contain muscle and that have been successfully replanted after 94 hours of cold ischemia.⁴⁵

Patient Evaluation

The amputated part is collected and appropriately packaged for transport with the patient to a replantation center. The amputated part is best preserved by wrapping it in gauze moistened with saline solution, placing this in a watertight plastic bag, and placing the plastic bag in a cooler filled with ice.⁴⁶ The amputated part should not be immersed in fluid and should not directly contact ice. The patient should be kept well hydrated, warm, and given analgesics for pain relief. In most cases, bleeding from the proximal stump can be controlled with direct pressure without a tourniquet. Clamping of vessels is unnecessary and should be avoided. On arrival at the replantation center, the patient and amputated part are examined, appropriate antibiotic and tetanus prophylaxis given, and radiographs of the stump and amputated parts obtained. Then a frank discussion with the child's family is of paramount importance to explain the predicted morbidity of replantation/revascularization, the prognosis for replant survival, the potential need for reoperation, and the anticipated functional outcome. We do not guarantee replant success and always discuss the possibility of completion amputation. Furthermore, we have found that allowing the child and parents to see the injury fosters better understanding of the extent of damage, improves overall rapport, and fosters acceptance of the final result.

Mechanism of Injury

The mechanism of injury significantly affects the potential for a successful surgical outcome. Guillotine-type amputations have a relatively small zone of injury and carry favorable results.⁴⁷ In contrast, degloving and avulsion-type injuries, at first sight, do not appear to have large zones of injury; however, closer inspection under an operating microscope inevitably reveals significant damage to the neurovascular structures.⁴⁸ Avulsion-type injuries are often identified by nerves hanging from the distal amputated stump and tendons that have avulsed from proximal muscle

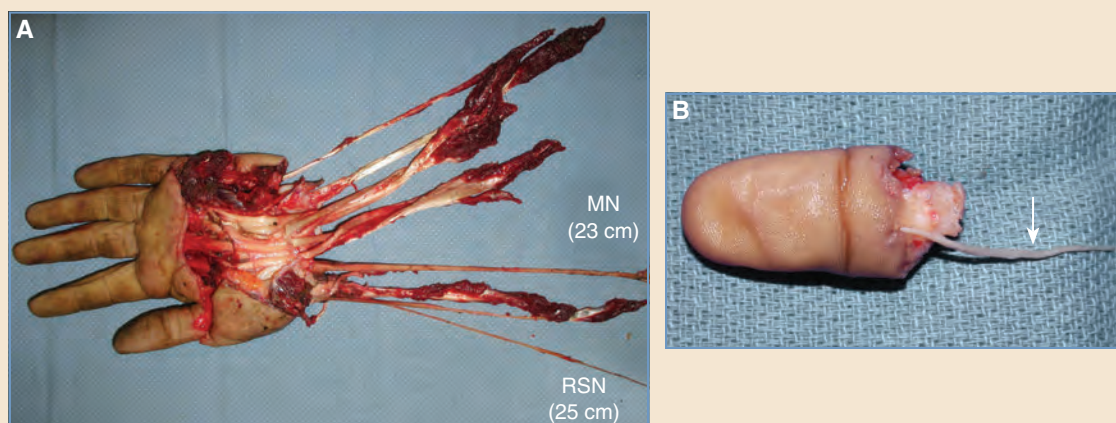


Fig. 52-26 **A**, A hand amputation showing tendon avulsions at the myotendinous junction with a 23 cm median nerve (*MN*) and 25 cm radial sensory nerve (*RSN*) avulsion. This hand was not replanted. **B**, An avulsion-type thumb amputation with a dangling digital nerve (*arrow*).

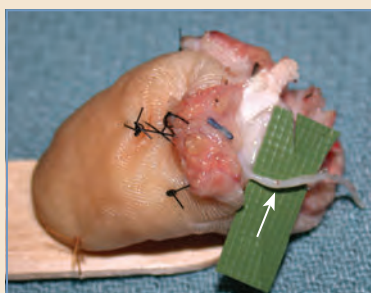


Fig. 52-27 A 10-year-old boy's thumb was amputated in a log-splitter accident. The palmar thumb is shown, with a palmar flap sutured back to expose the neurovascular structures. The amputated segment is stabilized on a sterile tongue blade. Before osteosynthesis, a forearm vein graft (*arrow*) is anastomosed.

bellies (Fig. 52-26). Crush injuries also carry a poor prognosis and have a large zone of injury. To help improve survival, aggressive debridement and vein grafting well out of the zone of injury are necessary.

Surgical Sequence

If possible, replantation should be performed using a two-team surgical approach. One team can take the amputated segment to the operating room (preferably) while the anesthesia team performs its workup. The part should be cleaned and debrided of nonviable tissues. In digital amputations, midlateral incisions are preferred for broad exposure of the vital structures. The digit can be sutured to a sterile tongue blade to serve as a stable platform for dissection (Fig. 52-27). Under an operating microscope the digital arteries, nerves, and dorsal veins are prepared for microsurgical anastomosis. We have found that tagging the arteries and veins with microhemoclips and the nerves with minihemoclips helps to readily identify these structures, particularly in a bloody field later in the operation. The tendons are tagged with sutures for future reference.

Before the induction of general anesthesia, a regional long-acting anesthetic block is given. Instead, preferably, an indwelling axillary catheter is placed for postoperative analgesia and vasodilation therapy. All bony prominences are padded, a Foley catheter is inserted, and warming

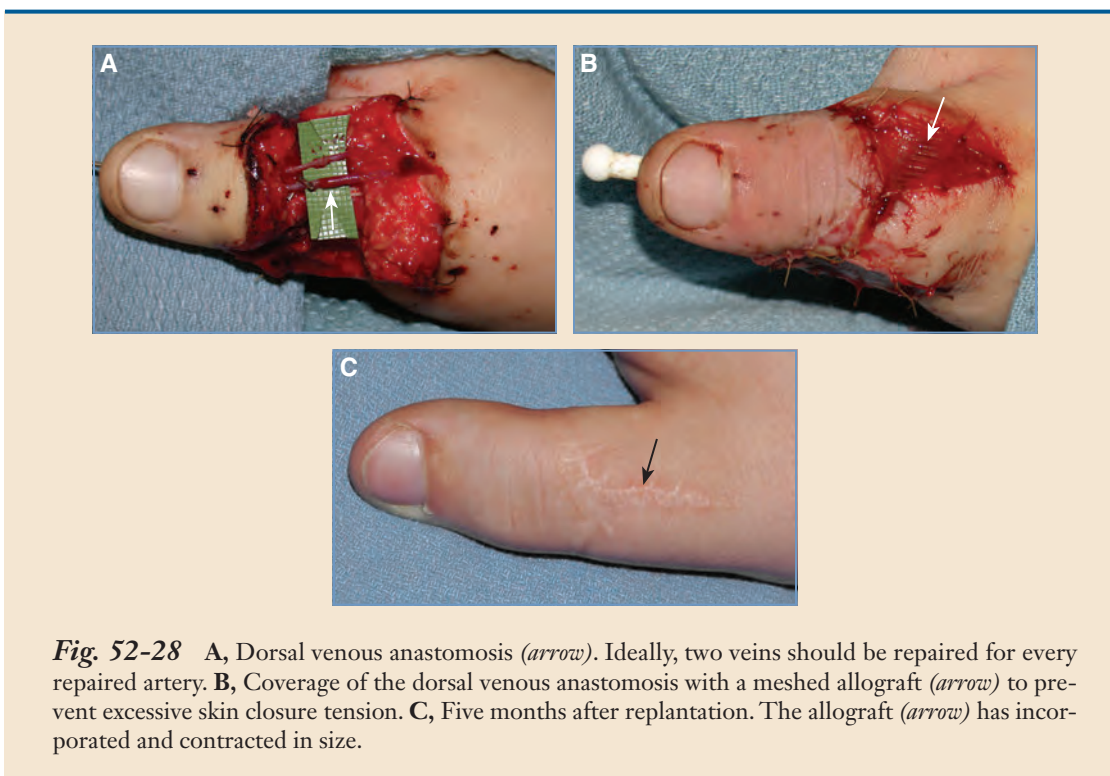
Box 52-2 Operative Sequence for Digital and Hand Replantation

1. Vessels, nerves, and tendons are identified and tagged.
2. Nonviable tissues are debrided.
3. The bone is shortened.
4. A stable bony fixation is obtained.
5. Tendons are repaired.
6. Nerves are repaired.
7. Readily identifiable veins are anastomosed; otherwise, arterial anastomosis is performed first.
8. The artery is anastomosed.
9. Skin coverage is completed.

blankets are used. At the outset of the operation, before exsanguination and tourniquet elevation, we routinely mark the course of the forearm veins in case vein grafts are later required. The typical operative sequence for digital and hand replantation is described in Box 52-2.

Several points deserve emphasis. Although its importance is often overlooked, obtaining rigid skeletal fixation allows early mobilization; thus it potentially facilitates superior postoperative range of motion. We have found that a 90-90 interosseous wire fixation technique provides rigid internal fixation and allows more aggressive postoperative therapy.⁴⁹ Another portion of the operation that is frequently performed in haste is the flexor tendon repair. Although the initial success of replantation and revascularization is judged by vascular patency, the ultimate result is related to the amount of active range of motion that is regained.⁵⁰ Therefore meticulous atraumatic technique is required for flexor tendon repair. We advocate using a 6-0 Prolene epitendinous suture, followed by a 4-0 nylon modified Kessler, and last a 4-0 nylon horizontal mattress suture.⁵⁰ The vascular repair deserves singular consideration. Under an operating microscope with high-power magnification (30×), the arteries and veins require careful inspection for intimal injury. Serial sectioning should be performed until there is no evidence of intimal damage. The vascular anastomosis should be tension free, and interposition-vein grafting should be readily performed to alleviate undue tension. We think that (if possible) at least two venous anastomoses should be performed (Fig. 52-28, *A*). The final step of this lengthy and delicate microsurgical operation is the skin closure. Inexperienced surgeons have a tendency to place excessive sutures when closing the wound. However, a tight closure leads to failure as the tissues swell and the thin-walled veins become compressed. The tissues need to be approximated loosely, and unmeshed skin grafts or allografts should be used liberally (Fig. 52-28, *B* and *C*). The use and mode of anticoagulants are controversial and the subject of debate. We give rectal aspirin just before the microvascular anastomosis and prescribe daily aspirin for 6 weeks. However, if the injury involves a significant crush component, in addition to aspirin, subtherapeutic intravenous heparin therapy is begun during the vascular anastomosis and is continued for 6 days.

In children, most amputations (unless the segment is severely damaged) should be replanted because of high overall rates of success,^{43,51,52} satisfactory return of sensation,⁵³ good total, active range of motion, and average bone growth of 93% compared with that of the uninjured side.^{53,54}



The superior regenerative capacity of children's nerves and soft tissues, along with the potentially favorable psychological ramifications of improved cosmesis, makes this technically demanding operation most gratifying.

ACUTE NERVE INJURIES

The hand functions as a receptor for pain, texture, and temperature and can provide a “seeing” function to discern the shapes of objects when the eyes are not used. Alterations in hand sensitivity will affect these functions, thus affecting the ability to perceive and manipulate the surrounding environment.

In children, upper extremity nerve injuries are caused mainly by laceration from sharp objects, such as broken glass, and to a lesser extent, by fractures (supracondylar humerus).⁵⁵ Neurologic and motor examinations are key factors in determining the presence and extent of nerve damage. However, these examinations can be daunting in younger children and in those who are painful and frightened. The neurologic examination should begin on the uninjured extremity to gain the patient's trust and to allow the patient to more clearly understand the examination process. The examination begins by subjectively examining the distal sensation to light touch using a cotton-tip applicator. A more sensitive method of assessing for acute nerve injuries is the moving two-point discrimination test, which objectively evaluates the quantity of innervated sensory receptors.^{56,57}

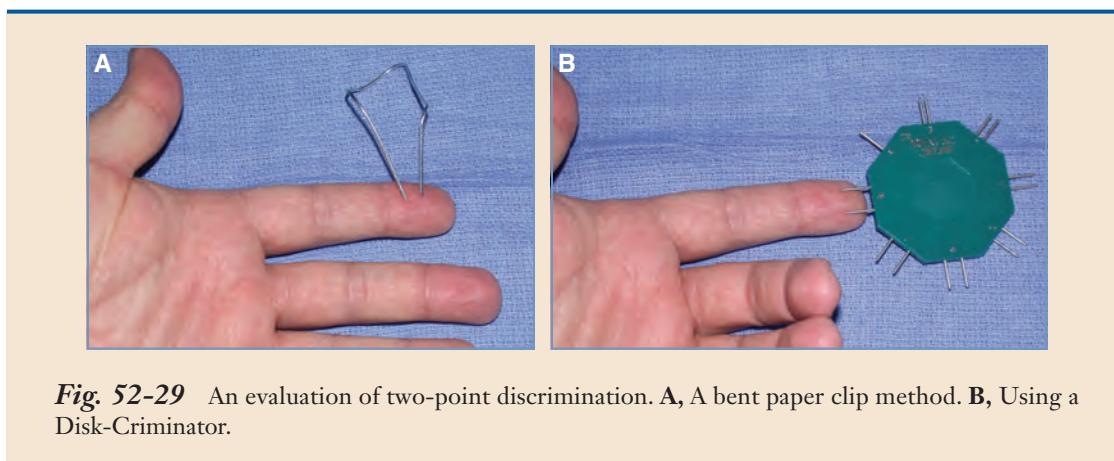


Fig. 52-29 An evaluation of two-point discrimination. **A**, A bent paper clip method. **B**, Using a Disk-Criminator.

The simplest method for performing this test is to use a bent paper clip⁵⁸; however, more reliable instruments are available⁵⁹ (Fig. 52-29). In infants and children with undeveloped or unreliable communication skills, the water immersion test can be useful to identify nerve injuries.⁶⁰ The hand is immersed in room-temperature water for 10 minutes. Sensate skin will wrinkle and “prune,” whereas insensate skin will not.

Classification of Nerve Injuries

When discussing nerve injuries, it is useful to classify injuries as a prognostic indicator of functional outcome. In 1943 Seddon⁶¹ proposed a classification system that is widely used today; it is based on the disrupted portion of the peripheral nerve’s internal structure. In 1968 Sunderland⁶² expanded on the Seddon classification system. This modified classification gave more emphasis to the injured fascicular layer.

Sunderland’s “first-degree injury” corresponds with Seddon’s “neuropraxia” and is a minimally demyelinating injury resulting in a temporary conduction block. Complete recovery can be expected in days to months. Sunderland’s “second-degree injury” is Seddon’s “axonotmesis,” involving Wallerian degeneration of the distal nerve. Because the endoneurial layer is intact, complete return of nerve function can be expected in a matter of months. Sunderland’s “third-degree injury” corresponds with Seddon’s “neurotmesis.” The endoneurium is affected and scars with this injury pattern; therefore recovery is mildly to moderately reduced. A fourth-degree injury is the more severe form of axonotmesis, and the endoneurium and perineurium are involved. Scarring of these layers can be significant, resulting in neuroma in continuity and moderate reduction of nerve function. A fifth-degree injury corresponds with neurotmesis. The peripheral nerve is completely severed, and no return of function is possible unless the nerve ends are surgically coapted.⁶⁰ Mackinnon and colleagues^{63,64} have described a sixth-degree injury—a mixed combination of various degrees of nerve injury.

Timing of Nerve Repair

Sharp nerve injuries should be explored and repaired immediately when possible. The greatest chance for an uncomplicated primary repair is before inflammation, scarring, and retraction of the nerve stumps has occurred. Delayed repairs have a higher chance of requiring secondary nerve

reconstruction procedures to span a nerve gap, for example, using nerve tubes, vein conduits, decellularized allograft, and autograft. Nerves that have been avulsed or crushed and those resulting from a blast mechanism of injury sometimes demarcate further over time. Some advocate for delayed debridement and reconstruction of the ensuing gap. However, we tend to perform the earliest possible debridement and repair/reconstruction possible within the context of the injury and other possible needs, such as vascular reconstruction, bony stabilization, contamination, tissue viability, and soft tissue reconstruction.

Once a nerve injury is diagnosed, the patient is taken for exploration and operative repair. A nerve repair performed within 24 hours of injury is called a *primary repair*. A repair done in the first week is called a *delayed primary repair*, and those carried out after 7 to 14 days are called *secondary repairs*. If conditions are optimal, nerve repair should be performed as soon as reasonably possible. In our experience, repairs within the first 24 hours are technically simpler, and primary and delayed primary repairs usually have the most favorable results.

Management

Optimal conditions for nerve repair include a sharply transected nerve that has minimal crush and avulsion components. The vascularized soft tissue coverage should be sufficient and the wound bed free of contamination. Indications for nonoperative management or delayed nerve repair include poor general patient condition, extensive soft tissue damage (Fig. 52-30), gross wound contamination, and extensive nerve damage for which tendon transfers will more rapidly restore function.

Several techniques are available for repairing injured peripheral nerves. Despite the controversy over which is the best technique,⁶⁵⁻⁶⁷ certain time-tested principles should be adhered to,^{68,69} including the following:

1. Motor and sensory function are assessed preoperatively.
2. Meticulous microsurgical technique is performed.
3. The repair is tension free.
4. An interposition nerve graft is used when tension-free repair is not possible.
5. If clinical conditions allow, a primary repair is performed.
6. The repair is delayed for 21 days if conditions are not optimal for primary repair.
7. Early, protected range of motion is initiated.
8. Occupational therapy is essential for sensory and motor reeducation.

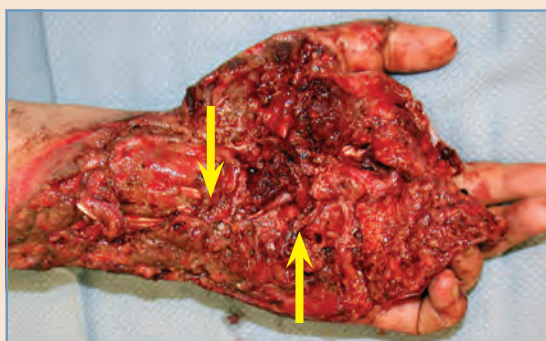


Fig. 52-30 This patient was involved in a rollover motor vehicle collision. The wound had extensive soft tissue damage, gross contamination, and segmental loss of the ulnar artery and nerve (arrows). The patient was managed with multiple debridements and soft tissue coverage, followed by delayed nerve graft reconstruction.

Techniques

End-to-end techniques are the preferred methods of nerve coaptation. Under an operating microscope, the nerve ends are serially sectioned until healthy, neuroma-free fascicles are encountered. The nerve is mobilized proximally and distally for a few centimeters to provide a tension-free repair. We consider tension to be appropriate when a single 8-0 or 9-0 nylon epineurial suture is capable of holding the nerve ends in approximation without pulling through. If the tension of the repair is deemed excessive, gentle joint-positioning maneuvers or nerve transposition can be performed. These maneuvers do not provide significant nerve stump advancement and are performed more to protect the repair itself if it is suspected of being under tension. If these maneuvers prove inadequate, the threshold for nerve grafting should be low.

An epineurial repair is considered the standard with which other repair techniques are compared. The fascicles are partially aligned using external topography (such as the longitudinal epineurial blood vessels) as a guide, and only the epineurium receives suture. This is a relatively fast technique that limits intraneurial trauma and is thought to have minimal foreign body reaction. Digital nerves receive two to three sutures and larger nerves receive eight to ten sutures of 9-0 nylon (Fig. 52-31), although the use of fibrin-based tissue glue can significantly reduce the number of nylon sutures needed. This technique is best suited for pure motor or sensory nerves and those with significant topographic crossover.

A group fascicular repair is used for larger nerves without topographic crossover, such as the distal median and ulnar nerves. This technique potentially improves repair accuracy by coapting matching fascicular groups with interfascicular perineurial suture. It is useful when performing cable grafts (Fig. 52-32).

A fascicular repair is a fairly uncommon technique that is used if individual fascicles are easily isolated. Each fascicle is repaired individually, providing more precise coaptation. The main disadvantages are that it is time consuming, the significant suture foreign body can result in intraneural scarring, and tension in the repair needs to be minimal. This technique is appropriate for repairing digital nerves distal to the trifurcation.

If tension on a nerve repair cannot be completely relieved by conservative means (for example, if the nerve gap exceeds 2 cm, and nerve mobilization and conservative joint positioning maneuvers are inadequate), then nerve reconstruction grafting should be performed. For short gaps, nerve tubes may suffice. The exact length of a nerve gap that will be well served by a nerve tube is not known but in some centers is up to 2 cm. We have much more limited indications for nerve tubes and will not typically use them for gaps larger than 5 to 10 mm. Instead, we opt for nerve grafting. Excellent sources for nerve grafts include the antebrachial cutaneous nerves, the sural nerve, and the posterior interosseous nerve for digital nerve repairs. The nerve grafts are interposed in the defect, and a fascicular repair is performed. The results of nerve grafting are better than repairs performed under tension. Alternatives to nerve grafting include processed nerve allografts⁷⁰ and nerve tubes or conduits made from varying materials, such as autologous vein,^{71,72} collagen, or polyglycolic acid⁷³; these have shown promising results in small defects.⁷⁴

Postoperatively, the patient's hand is immobilized for about 3 weeks, with only conservative joint flexion or extension maneuvers. Beginning on postoperative day 7, and on a weekly basis thereafter, the joint is slowly and incrementally brought to neutral position. Nerve regeneration



Fig. 52-31 Microscopic repair of a digital nerve with three 9-0 nylon sutures.

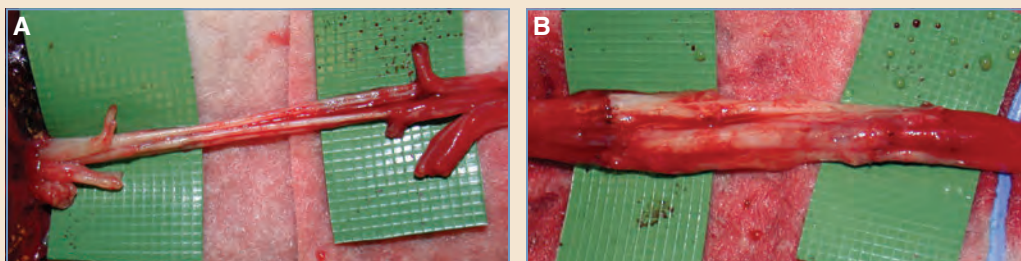


Fig. 52-32 **A**, A median nerve crush injury requiring neurolysis and neuroma excision. **B**, A group fascicular nerve graft repair of the median nerve.

halts for the first month after repair and resumes at a rate of 1 mm per day. The progression of regenerating nerves can be followed by an advancing Tinel sign. If no advancement occurs by 3 to 6 months, the repaired nerve should be reevaluated with nerve conduction studies or reexplored.

MUTILATING HAND INJURIES

A mutilated hand presents a significant challenge and is best cared for by hand surgeons proficient in reconstructive surgery and microsurgery. Caring for a child with mutilating hand injuries poses particular challenges with regard to more delicate anatomic structures, such as smaller-caliber vessels and tendons. The injuries may be less amenable to replantation and revascularization, because the mechanism of injury tends to be from meat grinders, saws, illegal fireworks,⁷⁵ and lawnmowers⁷⁶ (Fig. 52-33). To further complicate matters, parents often transfer their severe sense of guilt for not appropriately supervising their child to the health care team by being overly critical of the child's medical care or by becoming overly involved.⁷⁷ Despite these challenges, outcomes tend to be more favorable than in adults. Children inherently heal better and regain superior sensation with less long-term stiffness.

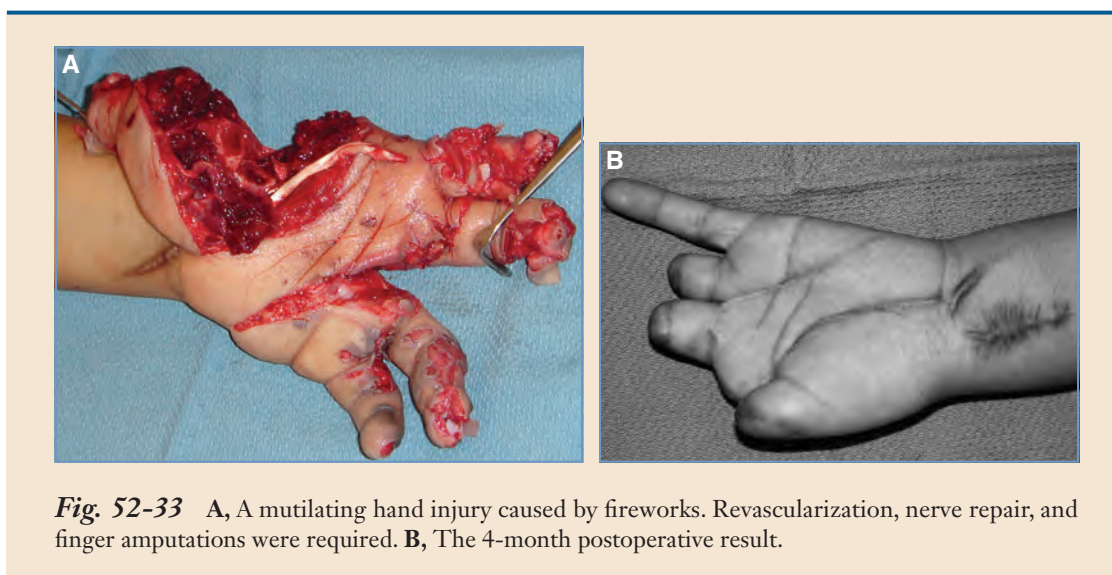


Fig. 52-33 A, A mutilating hand injury caused by fireworks. Revascularization, nerve repair, and finger amputations were required. B, The 4-month postoperative result.

The cornerstone of management of mutilating hand injuries involves early operative exploration, irrigation, and meticulous debridement of devitalized tissues. Care is required to prevent injury to vital structures such as tendons, nerves, and vessels during the debridement. We have found that copious pulse-lavage irrigation using saline solution impregnated with antibiotics aids in the safe debridement of foreign material and bacteria.

Damaged tissues are generally repaired in the same sequence as that used for a replantation, beginning with the larger, stabilizing structures such as bone osteosynthesis to maintain length, followed by tendon repairs.⁷⁶ Next, the neurovascular structures are repaired and the soft tissue coverage provided. After the arterial repair, the veins are allowed to bleed for several minutes before venous return is confirmed and before venous anastomosis to prevent the introduction of muscle breakdown products and proinflammatory mediators into the systemic circulation.

Although adequate soft tissue coverage is crucial for functional recovery after mutilating hand traumas, this coverage is often difficult to obtain. Flap coverage may be required; this should be delayed until the soft tissues are viable and infection free. To reach this stage, multiple trips to the operating room may be required for irrigation and debridement. In the interim, we have found negative pressure wound therapy to be an invaluable aid for decreasing tissue edema and for maintaining a moist environment over exposed tendons. Definitive soft tissue coverage depends on the defect and should be individualized for the patient. Wound coverage can be as simple as skin grafting or as complex as pedicled flaps and microvascular transfer. An ideal flap should provide vascularity and be thin and pliable, allowing appropriate joint movement, tendon excursion, and the potential for reelevation for revision surgery, including tenolysis/tendon reconstruction, delayed bone grafting, and hardware removal.

KEY POINTS

- Upper extremity injuries, particularly those of the hand, are common in toddlers as they explore their environment and in older children and teens who engage in sports.
- Differences in anatomy, growth potential, and rehabilitation outcome require adjustments to the treatment protocols used in adults.
- Closed reduction of Salter-Harris type II fractures is usually successful, but rotational deformities with scissoring cannot be left untreated.
- Flexor tendon repairs are performed much like those in adults except on a smaller scale, and postoperative protective immobilization is required in young children.
- Reconstruction requiring tenolysis or tendon grafting should be reserved for more cooperative older children and teens.
- Fingertip injuries are very common in toddlers, and careful, meticulous repair of bone and soft tissue can provide excellent results.
- Replantation of amputated parts is usually attempted in children as long as the part is suitable for replantation and prolonged surgery is not contraindicated.

ACKNOWLEDGMENT

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This group had a huge experience in devastating hand injuries. They were extremely innovative and creative in their reconstructive techniques. Their approach to management in both the acute phase and reconstructive phase is recommended.

53

Management of Obstetrical Brachial Plexus Palsy

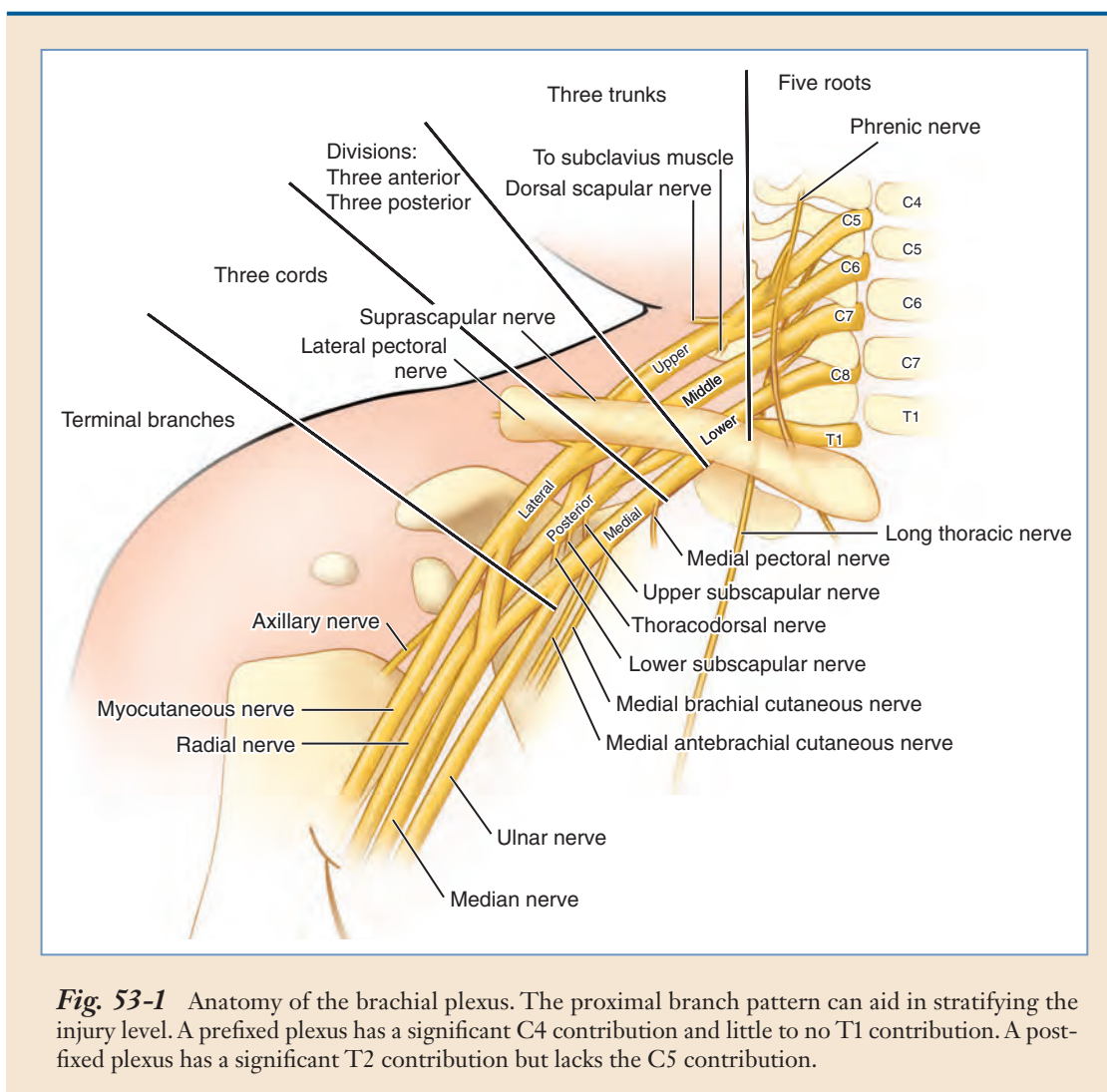
Kristen M. Davidge • Jeffrey R. Marcus • Howard M. Clarke



Obstetrical brachial plexus palsy is a traction neural injury that occurs during the birth process. As with all closed peripheral nerve injuries, the spectrum of severity is wide, and the degree of severity is the key determinant of prognosis and the need for intervention. Unlike single peripheral nerve injuries, the challenge of predicting severity and outcome for the brachial plexus is far greater because of the intricate pattern of this incompletely understood group of nerves. Opinions differ for managing obstetrical lesions of the brachial plexus in newborns. Microsurgical treatment in this area is a developing field. Surgeons engaged in early efforts to improve functional outcomes for infants with this potentially lifelong impairment quickly recognized the need to establish appropriate surgical criteria. Determining surgical indications is the most important and the most difficult step in caring for patients and families attempting to cope with the physical and emotional impacts of this condition. The factor most responsible for the disagreement among experts is the relatively incomplete understanding of the natural history of brachial plexus palsy. In this chapter we discuss the anatomy of the brachial plexus, the initial approach to patients with brachial plexus palsy, the natural history of the condition, the primary surgical treatment, and the expectations for outcome.

ANATOMY OF THE BRACHIAL PLEXUS

The anatomy of the brachial plexus is shown in Fig. 53-1. In most configurations the brachial plexus is formed from spinal roots C5-T1. *Prefixes* describes a plexus extending from C4, and *postfixes* describes a plexus extending to T2. As the roots emerge from their respective foramina, a characteristic branch pattern develops, which uses the following nomenclature: roots, trunks,



divisions, cords, terminal branches, and peripheral nerves. The trunks form as the roots converge between the anterior and middle scalene muscles. The upper trunk forms from C5 and C6, the middle trunk from C7, and the lower trunk from C8 and T1. Anterior and posterior divisions are formed for each trunk as they pass beneath the clavicle. Beneath the pectoralis minor the divisions converge with one another to form three cords: (1) the lateral, (2) the medial, and (3) the posterior cords, named with respect to their position relative to the axillary artery. The lateral cord is formed from the anterior divisions of the upper trunks, the medial cord is the continuation of the anterior division of the lower trunk, and all three posterior divisions form the posterior cord. Therefore the posterior cord contains input from all five of the spinal roots. Innervation of proximal muscle groups (that is, the shoulder girdle) originates from the root, trunk, and cord levels, whereas innervation of the upper and lower arm comes from terminal branches and ultimately from peripheral nerves more distally. The median nerve forms from contributions of the lateral and medial cords. The ulnar nerve and myocutaneous nerve are terminal branches of the medial and lateral cords, respectively. The radial and axillary nerves are terminal branches of the posterior cord.

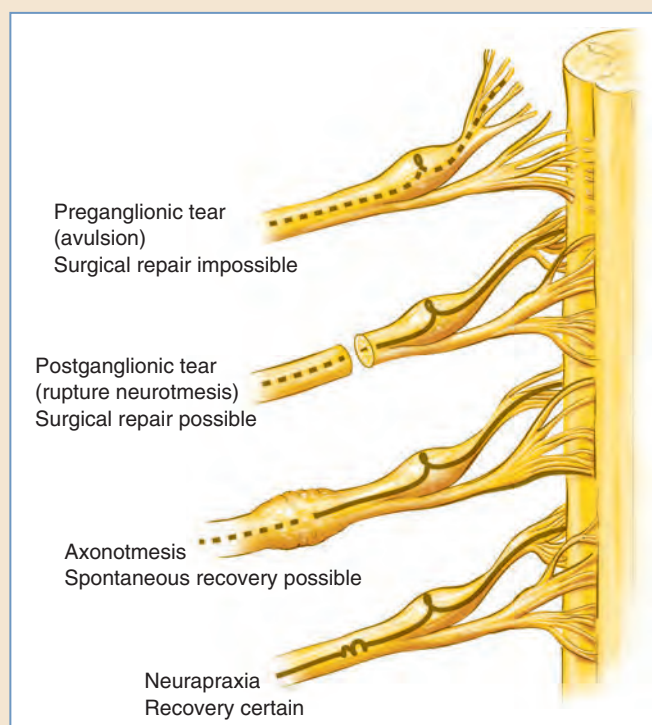


Fig. 53-2 Injury patterns to the spinal nerves. A preganglionic tear of the spinal nerve from the spinal cord is called an *avulsion*. Postganglionic tears are known as *ruptures*. A complete rupture is demonstrated by loss of continuity of the nerve. Also possible is an intraneural rupture of the fascicles in which some fascicles remain intact but others are dissociated within the epineurial envelope.

The level of injury within the plexus (from proximal to distal) can be determined to some extent from physical findings. A review of the brachial plexus anatomy—particularly the proximal branches—clarifies the location. For example, the presence of Horner syndrome indicates disruption of C8-T1 proximal to the sympathetic rami communicantes (a root avulsion). Scapular winging indicates a relatively proximal injury, because the long thoracic nerve supplying the serratus anterior originates at the root level distal to the foramina from C5, C6, and C7 contributions. The physical findings related to the level of injury are discussed in more detail in the discussion of physical examination.

Nerve injury patterns merit specific comment (Fig. 53-2). Injury of a single spinal nerve can occur at any level from the spinal cord origin to the terminal branches. The most proximal injury is a preganglionic tear of the spinal nerve rootlets from the cord, called *root avulsion*. Root repair, that is, replantation of avulsed rootlets, is currently not technically feasible. A postganglionic tear of the spinal nerve is a *rupture*. Whether the two free ends are separated, or whether they remain in continuity through only scar tissue, the axons are severed. Here, the proximal stumps are potential sources of viable axons for reconstruction, and the distal ends may serve as graft recipients. Intraneural rupture of fascicles results in a mixed injury; some fascicles remain intact and others are disrupted within the epineurial envelope. The result—neuroma in continuity—is the most frequent finding at surgery for brachial plexus reconstruction. Obstetrical injuries to the brachial plexus can result in variable combinations of all of these injuries.

EPIDEMIOLOGY

In modern obstetrical practice, the incidence of obstetrical brachial plexus injury is estimated at 0.5 to 2 per 1000 births.^{1,2} Included in this estimate are those who progress quickly to complete recovery and those who improve slowly and incompletely. In reported series, the rate of complete, spontaneous recovery varies widely (30% to 95%).³⁻⁷ Lower reported rates reflect, in part, preselection; those who recover quickly and completely often are not referred to specialists and may be excluded from analyses. The definition of *recovery* is inconsistent. Although many infants with plexopathy recover with minor or no residual functional deficits, some children do not regain sufficient limb function and develop functional limitations, bony deformities, and joint contractures. Imbalanced shoulder girdle musculature resulting from incomplete recovery can lead to subluxation of the shoulder and deformation of the glenoid.^{8,9} Bony deformities and contracture can occur even in cases with reported complete neurologic recovery.¹⁰ Birch^{11,12} stated that palliative procedures for secondary deformities are deeply unsatisfactory, and the results of myotendinous transfers in obstetrical palsy are far inferior to those that follow good nerve regeneration. The challenge of statistical prognostication is to identify children most likely to benefit from exploration and reconstruction of the plexus.

HISTORY

At presentation, a detailed history should be taken that addresses maternal factors, the prenatal course, events of the delivery, and the postnatal course. Fig. 53-3 presents an example of the historical data reference used at our institution. For the purposes of data collection, historical items in this format have been separated into two categories: maternal factors and child factors. Maternal factors include data pertaining to the prenatal history, such as maternal diabetes, preeclampsia, duration of labor, and previous obstetrical history. The natal history described by the parents includes delivery presentation, the use of instrument assistance, and shoulder dystocia. Child factors include gestational age, weight, and complications such as respiratory complications, fractures, diaphragm paralysis, and shoulder subluxation.

Historical data are documented for the purposes of population study. Some centers that receive a larger number of referrals have an internal database that allows analysis and reporting of relatively large series, which has led to our current, yet still incomplete, understanding of the disease process. Participation of all centers, particularly smaller ones, in web-based, multicenter data collection programs may be helpful. One example is the multicenter database sponsored by Children's Hospital of Boston. This database compiles historical information and serial examination data. With regard to history, attention is focused on factors that have reported statistical associations with obstetrical brachial plexus injury. The factors of interest are those with suspected causal relationships to the injury, those that might indicate a high level of energy at the time of injury, and those that may suggest the anatomic level of injury.

Several associations have been consistent in the literature. The average birth weight of affected infants is reported to be 500 to 1000 g heavier than that of unaffected infants.¹ The reported association of maternal diabetes with obstetrical lesions is probably a result of its effect on birth weight. After the first pregnancy, birth weights tend to increase with each successive pregnancy. Fetal macrosomia is another condition that, although cited separately, is associated with higher average birth weight.¹³ Generally, these conditions share the same potential delivery-related difficulty, that is, cephalopelvic disproportion.^{14,15} Shoulder dystocia is perhaps the most common association, and the mechanism of vertex brachial plexus injury is most easily understood in this context. It is generally accepted that injury to the brachial plexus is a result of lateral displacement of an infant's head away from the shoulder during the last phase of delivery.^{1,14,16,17} Other

THE HOSPITAL FOR SICK CHILDREN'S OBSTETRICAL BRACHIAL PLEXUS LESIONS DATABASE

Referring Physician _____

Age: _____ Affected side: ☐ Right ☐ Left

Maternal History

Prenatal

Diabetes: ☐ Yes ☐ No

Duration of labor: _____

Preeclampsia: ☐ Yes ☐ No

Parity/Gravida: ☐ P ☐ G

Delivery problems with earlier pregnancy: ☐ Yes ☐ No

Natal

Birth center: _____

Position: ☐ Vertex ☐ Breech ☐ Other

Delivery: Uncomplicated/difficult: _____

Normal/forceps/vacuum/C-section: _____

Dystocia: Head/shoulder: _____ Right/left/both: _____

Child Factors

Large baby: ☐ Yes ☐ No

Birth weight: _____ Gestational age: _____

Complications

Asphyxia: ☐ Yes ☐ No

Respiratory complications: ☐ Yes ☐ No

Clavicle fracture: ☐ Yes ☐ No

Horner syndrome: ☐ Yes ☐ No

Rib fracture: ☐ Yes ☐ No

Hemidiaphragm paralysis: ☐ Yes ☐ No

Humerus fracture: ☐ Yes ☐ No

Shoulder subluxation: ☐ Yes ☐ No

Torticollis: Left/right/none: _____ True/positional: _____

Mild/moderate/severe: _____

Fig. 53-3 An example of a data sheet for collecting historical information.

reported associations include brachial plexus injury in previous pregnancies and the use of vacuum or forceps assistance during delivery.

The incidence of concurrent delivery-related injuries, such as fractures or dislocations, needs to be determined. Some think that such injuries provide meaningful clinical suggestions regarding the applied energy at the time of injury. For example, a fracture of the humerus may indicate significant applied energy. Humeral fractures may also be associated with pseudoparalysis, in which neurologic impairment occurs as a result of direct impingement of the plexus by the fracture itself. Clavicular fractures have been associated with obstetrical plexus injury,⁶ but some suggest that it may actually be protective to a certain degree,¹⁷ allowing downward rotation of the shoulder-scapular complex during dystocia. Shoulder dislocations can occur with or without injury to the brachial plexus. The literature does not provide a statistical association of prognosis with the incidence of humeral and clavicular fractures.¹⁸

Brachial plexus injuries are most frequently reported after vertex deliveries, but they have also been reported with breech deliveries and cesarean sections. Metaizeau et al¹⁷ investigated the effects of traction on the brachial plexus in stillborn infants. Immobilizing the shoulder and applying lateral traction to the head demonstrated force requirements and patterns of injury. Epineurial ruptures occurred at a lower force level than complete ruptures. Ruptures were more common in the upper roots and avulsions in the lower roots.¹⁶ The findings of Metaizeau et al¹⁷ coincide with clinical observations and injury patterns in vertex deliveries with plexopathy. In breech deliveries with plexopathy, a lower-than-normal birth weight is more common, whereas the converse is true with vertex deliveries. The characteristic injury pattern tends to be more severe in breech deliveries, particularly at the upper roots. Avulsions of C5 and C6 are frequently seen.¹⁹⁻²¹ Brachial plexus injuries with breech presentations have been most clearly documented by European groups. Recently Al-Qattan²² reported on a series of patients with obstetrical plexopathy in association with breech delivery in Saudi Arabia, which echoes the findings noted previously. Relatively fewer cases of breech-related plexopathy are seen in North America, perhaps because of a greater tendency to perform a cesarean section under such circumstances. However, investigators have noted an approximately 1.5% incidence of brachial plexopathy after cesarean section.^{5,16,23} Clinical evidence of brachial plexus trauma with characteristic neuroma formation is described for reported postcesarean patients who proceed to exploratory operations. An urgent cesarean section may follow a difficult initial attempt at vaginal delivery. In such cases, the same potential injury mechanisms resulting from vertex vaginal delivery may occur. The perceived inconsistency of injury after cesarean sections with the commonly accepted mechanism for vertex-related plexopathy has prompted some to suggest the possibility of intrauterine injury to the plexus or congenital malformation. This hypothesis is not yet supported by conclusive data.

PHYSICAL EXAMINATION AND MOTOR ASSESSMENT

A thorough examination should be conducted, with the following goals:

- Determining the presenting functional severity
- Identifying other traumatic injuries or complicating factors
- Querying for possible confounding diagnoses

Physical Examination

The head, neck, and shoulders are examined. The position of the head is assessed for favored laterality, which could indicate the presence of either primary or secondary torticollis. Primary (true) torticollis can be seen immediately after birth in infants with a brachial plexus injury and after an uneventful delivery in infants without brachial plexopathy. However, children with



Fig. 53-4 The typical posture of an infant with right upper trunk palsy. The shoulder is held in adduction with the elbow straight. The wrist, fingers, and thumb are flexed. This is sometimes referred to as the *waiter's tip position*.

brachial plexus injury tend to look away from the affected side despite the absence of true torticollis. Parents tend to nurse with the child's affected arm up. Unless the condition is diagnosed and treated with physical therapy, sternocleidomastoid shortening and secondary torticollis can gradually develop.^{16,23}

The upper extremities are carefully examined with the child at rest. After a brachial plexus injury, one of a few distinct patterns is followed. The first is characteristic of upper plexus palsy (Erb palsy), in which deficits are primarily at the shoulder and elbow. The level of involvement in this injury is C5-6, and possibly C7. The resting position, which favors the more richly innervated or least affected muscle groups, is characterized in upper root palsy by adduction and internal rotation of the shoulder, extension of the elbow, pronation of the forearm, and flexion of the wrist and fingers. This position is sometimes referred to as the *waiter's tip position* (Fig. 53-4). A second common injury pattern is total plexus palsy, which is characterized by complete atonia of the upper extremity (C5-8 and possibly T1). These infants often give little to no attention to the affected extremity. Klumpke paralysis, or lower plexus palsy, refers to isolated involvement of C8 and T1, and it is generally not seen as a manifestation of obstetrical palsy.²⁴ The pathology of intermediate plexus palsy, primarily affecting C7 and sometimes C8 and T1, is well described.²⁵ However, it is generally not characterized by a unique or classic position at rest during the early period of evaluation. The appearance usually resembles that of upper or total plexus palsy, depending on the extent of involvement of the less injured roots. A final injury pattern, isolated C7 avulsion, is uncommon and presents primarily as a flexion deformity at the elbow.

The clavicles, humeri, and ribs are evaluated for fractures. The position and posture of the shoulders are evaluated for dislocation. The scapulas are difficult, if not impossible, to evaluate in infants. Asymmetry and winging in older children may suggest a proximal injury with impairment of the long thoracic nerve. The phrenic nerve is another proximally based branch, with contribution from C3-5.^{26,27} Breathing is carefully evaluated to determine involvement of the phrenic nerve. Symmetrical movement of the rib cage with coordinated abdominal movement should be seen. In our practice, all preoperative patients receive ultrasound imaging of the diaphragm. Postoperatively, in some cases, transient hemiparalysis of the diaphragm can result from traction during the course of dissection. If such a patient had a normal preoperative study, resolution is

probable with expectant management. In principle, a paralyzed hemidiaphragm may require plication to improve function; however, we have not encountered this situation. Finally, the pupils and eyes are examined for signs of Horner syndrome: ptosis, miosis, anhidrosis, and enophthalmos. These findings suggest a more proximal injury^{19,28,29}; specifically, they indicate disruption of T1 proximal to the sympathetic rami communicantes. Preganglionic sympathetic fibers arise from T1 (and often T2). Just after T1 exits its intervertebral foramen, the fibers branch from the spinal nerve to enter the sympathetic chain through the white rami communicantes. They subsequently travel to the superior cervical ganglion and synapse. From there, postganglionic fibers proceed along the carotid plexus to innervate the dilator pupillae muscle and levator palpebrae muscle. A T1 injury distal to the rami communicantes does not disrupt the sympathetic fibers; therefore it does not result in Horner syndrome.

Motor Assessment

Motor assessment is perhaps the most important component of a systematic examination. In neonates and infants, however, it is the most difficult. Accuracy and consistency require patience and experience and are important for decision-making for each patient and for systematic evaluation of global treatment plans. Assessment of motor function in older children and adults is more straightforward and involves the use of grading scales based on a level of demonstrable voluntary power. The British Medical Research Council's familiar 0 through 5 grading scale is shown in Table 53-1.³⁰ Because infants cannot participate in testing that requires voluntary demonstration of full power, some investigators have sought to develop instruments based on range of motion rather than power. Such is the case for the muscle grading system of Gilbert and Tassin³¹ (Table 53-2), which considers the presence or absence of movement with and without the effect of gravity. The scale is simple, easy to apply, and widely recognized. However, because it must cover a wide range of ability within four possible grades (M0 to M3), its discriminatory capacity is limited. Furthermore, the scale has never been subjected to validation studies.

The Hospital for Sick Children's Active Movement Scale is an eight-point assessment scale for active range of motion^{16,32} (Table 53-3). It was designed to be highly discriminatory, capturing subtle and significant changes in movement.^{16,23} Similar to the grading system of Gilbert and Tassin,³¹ it addresses range of motion in the presence and absence of gravity. Further, however, it quantifies the active range under each of these conditions. A number of guidelines have been

Table 53-1 British Medical Research Council's Muscle Grading System

Observation	Muscle Grade
No contraction	0
Flicker or trace of contraction	1
Active movement with gravity eliminated	2
Active movement against gravity	3
Active movement against gravity and resistance	4
Normal power	5

Data from British Medical Research Council, *Aids to the Investigation of Peripheral Nerve Injuries*. London: His Majesty's Stationery Office, 1943.

Table 53-2 Gilbert and Tassin's Muscle Grading System

Observation	Muscle Grade
No contraction	M0
Contraction with movement	M1
Slight or complete movement with weight eliminated	M2
Complete movement against the weight of the corresponding segment of the extremity	M3

Data from Gilbert A, Tassin JL. Obstetric palsy: a clinical, pathologic and surgical review. In Terzis JK, ed. Microreconstruction of Nerve Injuries. Philadelphia: WB Saunders, 1987.

Table 53-3 Hospital for Sick Children's Active Movement Scale

Observation	Muscle Grade
Gravity Eliminated	
No contraction	0
Contraction, no motion	1
Motion $\leq \frac{1}{2}$ range	2
Motion $> \frac{1}{2}$ range	3
Full motion	4
Against Gravity	
Motion $\leq \frac{1}{2}$ range	5
Motion $> \frac{1}{2}$ range	6
Full motion	7

Data from Clarke HM, Curtis CG. An approach to obstetrical brachial plexus injuries. Hand Clin 11:563; discussion 580, 1995.

developed in an effort to instruct others in its use and to standardize application of the scale. For example, infants often demonstrate limited movement both with and without gravity effects. With the Active Movement Scale, infants cannot be scored against gravity unless they first achieve a score of 4, reflecting a full, active range without gravity. Gravity-eliminated movements are always assessed first to determine whether higher scores might subsequently be assigned. The scoring system is applied to the key movements of each potentially affected joint: shoulder flexion, abduction, adduction, internal and external rotation, elbow flexion and extension, pronation and supination of the forearm, wrist flexion and extension, finger flexion and extension, and thumb flexion and extension. Practical applications of the instrument have been described in detail in the literature^{16,23,32}; the specific guidelines and suggestions are essential to its consistency. Reliability studies of the Active Movement Scale have shown consistent intrarater reproducibility and interrater reliability among experienced and recently trained physiotherapists. Variability is a result of patient factors rather than rater factors.³²

Here is a final note on the motor examination: It is just as important for any grading scale to be reliable and discriminatory as it is to be applicable along the full range of a child's development.

	II	III	IV
ACTIVE ABDUCTION	 Inferior to 30°	 30° to 90°	 Superior to 90°
EXTERNAL ROTATION	 0°	 Inferior to 20°	 Superior to 20°
HAND TO NAPE OF NECK	 Impossible	 Difficult	 Easy
HAND TO BACK	 Impossible	 S1	 T12
HAND TO MOUTH	 Claron	 Small claron	

Fig. 53-5 Mallet's classification system evaluates global movement of the extremity to identify functional or maladaptive patterns. It requires cooperation from the patient to perform voluntary movements on command.

A major advantage of scales such as the Active Movement Scale and Gilbert and Tassin's muscle grading system³¹ is that they can be administered during infancy and repeatedly throughout development, including postoperatively. This allows consistent analysis of the patient and is a statistically sound means to study the natural history of the injury and postoperative results. In 1972 Mallet^{33,34} established a grading scale that addressed global movement of the extremity and functional adaptations³⁵ (Fig. 53-5). It is widely used and assesses the functional utility of the extremity by grading such composite movements as "hand to nape of neck." Mallet's classification^{33,34} is limited because directed, complex movements are not reliably provoked in infants, who often are first examined at 1 to 3 months of age. We strongly support consecutive application of the same test instrument during development for both clinical and analytic purposes; therefore we have not incorporated the Mallet classification into our routine clinical practice.

PROGNOSIS AND THE DECISION FOR SURGICAL EXPLORATION

Most investigators agree that children who will ultimately require surgery should be treated as early as possible to maximize the benefit of the intervention. The difficulty lies in accurately identifying these children early. There are two circumstances in which, given our current level

of understanding, the indications for surgery are quite clear early on. First, as Waters³⁶ indicated, normal neurologic function can be almost universally expected without surgery in patients who demonstrate recovery of neurologic function within 1 month after birth. His conclusions, which support the work of Eng et al,¹ Michelow et al,⁵ and Tassin,³⁷ were based on the first noted appearance of biceps function by 1 month in 8 of 66 patients who were followed for a minimum of 2 years. A large proportion of patients probably fall into this group. Although we would prefer to follow all patients, realistically many patients in this category may recover without ever proceeding to a specialist referral. At the opposite end of the spectrum, a poor, long-term functional result is expected if no intervention is undertaken for an infant who has total plexus palsy with signs of Horner syndrome and fails to improve over the first 3 months. Numerous investigators have stated that the presence of unequivocal Horner signs inevitably indicate a need for early exploration.^{16,28,29} In Waters' series,³⁶ 8 of 66 patients (12%) demonstrated recovery within 1 month; only 3 of 66 (5%) had total palsy with Horner syndrome. Thus nearly 80% of the patients in this tertiary referral-based series presented with injury severity between these extremes, indicating that additional data and selection criteria are necessary.

Surgery is indicated if evidence shows that the outcome after intervention is likely to result in better long-term function than that resulting from conservative management. To make this judgment, the natural history of the condition must be known along its full spectrum of severity. If predictors of poor outcome can be identified and used as surgical criteria, then a comparative outcome study could be conducted to evaluate treatment effects. The approach taken by Tassin³⁷ and Gilbert and Tassin³⁸ in 1984 focused on predicting shoulder outcome in 44 patients with obstetrical palsy. They observed that over a 5-year period, a "good shoulder" (Mallet class IV) was not obtained unless biceps and deltoid contraction began by the third month.^{37,38} They reported, "An average (class III) or bad (class II) shoulder developed if biceps or deltoid recovery began after the third month." Because of the perceived difficulty in evaluating deltoid function, the authors recommended the absence of elbow flexion alone at 3 months as the primary indication for surgical exploration. The absence of elbow flexion at 3 months has become the most widely accepted criterion for surgery in cases that fall between the two previously mentioned extremes (i.e., normal or no neurologic recovery).

Some think this criterion may result in a too-high false-positive rate and a surgical recommendation for some children who would otherwise recover satisfactorily with nonoperative treatment. In our study of the natural history, we reported the progressive changes in active movement over a 12-month period in 66 patients with obstetrical palsy.⁵ In this series, patients were evaluated repeatedly at 6-month intervals after birth using an earlier version of the Hospital for Sick Children Active Movement Scale. The cardinal movements at the shoulder, elbow, forearm, wrist, and hand were graded. At 12 months, patient outcomes were assessed according to the Narakas classification,³⁹ in which poor recovery was defined as elbow flexion of half or less of the normal range and shoulder abduction of less than half the normal range. For a functional range beyond these limits, recovery is considered to be good. We conducted an analysis to determine whether the recovery of any one motion or set of motions at 3 months could statistically predict poor recovery. Absent elbow flexion alone at 3 months incorrectly predicted a poor recovery 12.3% of the time. Using a composite score of elbow flexion plus elbow, wrist, thumb, and finger extension at 3 months, poor recovery was incorrectly predicted 5.2% of the time at a cutoff score of 3.5. Therefore we concluded that an additional 7.1% of children could be spared surgery and still be expected to achieve a good recovery if the more comprehensive score is used.

Waters³⁶ also investigated recovery of elbow flexion as an independent criterion for surgery. In his very elegant and careful study of 66 patients with obstetrical palsy, he evaluated recovery of biceps function monthly from birth to 6 months of age. The outcome was based on the Mallet global functional movement scale. Mallet scores were determined at each evaluation, and the minimum follow-up for all patients was 2 years. Waters found that patients who recovered biceps

function within 4 to 6 months after birth did not proceed to normal global function, unlike those who recovered biceps function within the first 3 months. For those who underwent surgery at 6 months because of biceps recovery failure, the outcome was better than for those who regained function at 5 months and did not undergo surgery. The findings further support the idea that the specific timing of recovery is an important prognostic variable, and that surgical outcome can surpass natural history in carefully selected patients. Therefore Waters concluded that, in addition to patients with total palsy who do not improve by 3 months, surgery should be offered to patients who do not recover biceps function by 5 months.

Strombeck et al² studied 247 patients, of whom 54% recovered function of the biceps within 3 months and later progressed to satisfactory function. They reported unchanging total plexus palsy at 3 months in 13% of patients and recommended surgery for all of them. Most of the remaining patients (33%) were observed for “late recovery,” which was defined as biceps recovery at the 6-month or 9-month evaluation. Those who did not regain biceps function at these later times were recommended for surgery. The authors found that late recovery occurred, and no significant difference was noted in outcome between the group that underwent surgery and the group that did not, except for improved shoulder movement in those with upper plexus lesions. The findings led the authors to reject the 3-month biceps recovery criteria altogether in favor of waiting for late recovery. Although we agree that a small number of patients regain biceps function after 3 months and proceed to satisfactory recovery, waiting for late recovery in all cases is not appropriate. In our natural history study, half of the patients with no elbow flexion at 3 months later developed good function, but the other half did not. The test score developed from this study was designed to identify patients with a statistical likelihood of poor recovery while limiting the number of incorrect predictions. Based on this information, an early surgical recommendation may have been given to some of the patients in the Strombeck study. Although the surgical patients in their study (those with more severe injury) had later operations, they still had functional recovery that was at least as good as the recovery of those who were observed (those with presumably less severe injury). A careful statistical approach to surgical indications, such as those of Waters³⁶ or Michelow et al,⁵ can limit low-yield interventions while maximizing the potential benefits of surgery by making early recommendations when appropriate.

The false-negative findings with the elbow flexion criteria of Tassin³⁷ and Gilbert and Tassin³⁸ at 3 months are a concern. In cases with a normal test result (flexion of the elbow), surgery would be deferred. A small percentage of children who meet such criteria at the 3-month interval may still have a poor recovery. Therefore assessments must continue to be carried out at 3-month intervals to ensure that children with early evidence of recovery progress satisfactorily.

In our practice, a test score is determined at the 3-month assessment based on the following statistical discriminators, which together were shown to limit the false prediction rate for poor recovery to only 5.3%: elbow flexion and elbow, wrist, finger, and thumb extension.⁵ Patients with a test score of greater than 3.5 are observed and continue physical therapy to maintain their range of motion. Active Movement Scale scores are assessed every 3 months. Some patients with upper trunk lesions who show good early recovery and have test scores greater than 3.5 may not develop adequate elbow flexion by the end of their first year and may have poor shoulder function. In these cases, surgical exploration is offered for patients who at 9 months of age demonstrate elbow flexion of less than grade 6 (less than half the range of motion against gravity). At this age children can better cooperate during evaluations of more complex functional movements; therefore elbow flexion is assessed with what we call the *cookie test*. The elbow is held in adduction at the trunk, a small cookie is placed in the child's hand, and the child is encouraged to bring it to his or her mouth. The child passes the test and is rejected as a surgical candidate only if he or she cannot take the cookie to the mouth by elbow flexion against gravity and with less than 45 degrees of neck flexion.^{16,32}

Surgery is recommended for patients who do not achieve a test score of 3.5 at the 3-month examination (capturing the most severe cases) and for patients who fail the cookie test at the 9-month interval (capturing the mildest cases that will benefit from surgery). For some children, surgery is offered at the 6-month interval if they initially demonstrated elbow flexion at the 3-month assessment but failed to progress. If they have not progressed since the first few months and the 3- and 6-month scores are similar, further improvement is unlikely. At 6 months, infants generally are not yet able to cooperate with the demands of the cookie test. Insufficient satisfactory progress in test scores between 3 and 6 months of age serves to identify intermediate cases. These children might have benefited from surgery at an earlier time.

Some infants pass the cookie test at 9 months of age but do not recover good shoulder function. We recently revised the Hospital for Sick Children algorithm for evaluation and management of infants with obstetrical brachial plexus palsy to include inadequate spontaneous recovery of shoulder function as a separate indication for a primary nerve operation at 9 months of age⁴⁰ (Fig. 53-6). This change in practice was based on a review of 17 patients who were offered

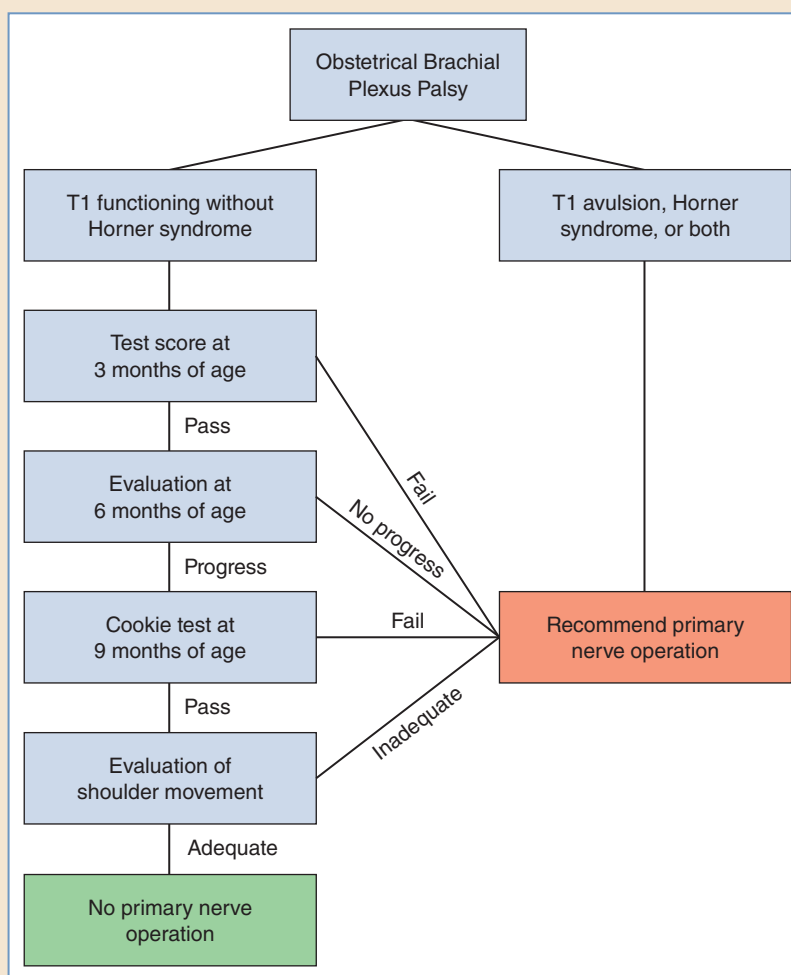


Fig. 53-6 A flow diagram of the extended indications for primary nerve surgery in obstetrical brachial plexus injury used in our center.

a primary nerve operation between 2004 and 2009 on the basis of poor shoulder function, despite adequate elbow flexion.³⁷ Sixteen of these patients had no active external rotation (Active Movement Scale score of 0), and, of more importance, all patients had sufficient passive range of motion to demonstrate poor active range. Of the 14 patients who underwent surgery, 11 patients had an isolated suprascapular to spinal accessory nerve transfer from an anterior approach for absent active external rotation. The other 3 patients underwent a neuroma excision and grafting of the upper trunk, or upper and middle trunk, because of poor active range in shoulder flexion and abduction. All patients in the operated group regained some active external rotation of the shoulder (Active Movement Scale scores ranging from 2 to 7), and they regained or surpassed their scores for other shoulder motions. By contrast, in the nonoperated group, 2 of 3 patients did not demonstrate further recovery in active external rotation, and it remained absent.

All appropriate patients should be offered surgical reconstruction of brachial plexus palsy lesions. For children most severely affected, this decision is made when they are 3 months of age; a failure of the test score, a positive Horner sign, or clinical evidence of a T1 avulsion is sufficient to indicate that surgical reconstruction should be offered. At the other end of the spectrum, a failed cookie test or poor shoulder function in children 9 months of age indicates some degree of early spontaneous recovery but failure to reach an adequate plateau that would suggest a good final outcome. Patients with an intermediate pattern are treated at an intermediate time interval.

The decision to perform surgical exploration of the brachial plexus in infants is not easily made. Challenging questions about management do not end at this point. Exploration of the brachial plexus is a technically elegant first stage of the operation, exposing and revealing the grossly apparent sequelae of a closed traction injury. Once the plexus is exposed, more challenges arise, including identifying root avulsions and managing neuroma-in-continuity lesions by neurolysis, neuroma resection and grafting, or neurotization.

SURGICAL APPROACH: EXPLORATION OF THE BRACHIAL PLEXUS

Positioning

The infant is placed in the supine position fully at the head of the bed and close to the lateral edge on the injured side (Fig. 53-7). This allows the surgeon and assistants to work from above, beside, and below the field. The entire upper extremity, upper chest, and neck are prepared in the field. The arm is placed in a towel, which is clipped to the drape at waist level. The arm is internally rotated with the elbow flexed to 90 degrees for most of the procedure. It will be accessible for examination when the exposed plexus needs to be stimulated. The head is turned to face away from the injured side. A small sandbag is placed beneath the shoulder to provide an appropriate amount of neck extension. All pressure points, particularly on the scalp, are meticulously padded. Our surgical team and anesthesiologists favor securing a nasotracheal tube with a suture, and we routinely drape the head using transparent material to facilitate monitoring the airway throughout the lengthy operation. A rectal temperature probe is used, and appropriate standard techniques for temperature maintenance are employed with vigilance.

Incision

Among experienced plexus surgeons, the choice of incision varies. Some choose to start with a single transverse incision.³⁹ We prefer to raise a V-shaped flap for complete exposure of the posterior triangle; the incision follows the posterior border of the sternocleidomastoid, gently curves just above the clavicle, and follows a line parallel to the clavicle (Fig. 53-8). This incision provides optimal exposure of the entire field of interest and prevents the need for adjustments and shifts throughout the procedure that are necessary with a transverse incision. Occasionally,

Fig. 53-7 The infant is positioned to allow the surgeon to work above, beside, and below the field. The neck is slightly extended, and the head faces away from the field. The hand and arm are accessible for observation of stimulated movements. A clear plastic drape covers the patient's face and the nasotracheal tube so that they are always visible to the anesthetist and the other members of the surgical team.

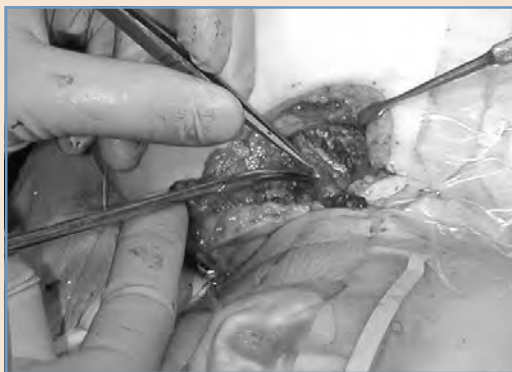


Fig. 53-8 An incision is made along the posterior border of the sternocleidomastoid and the clavicle, and a V-shaped flap is created and reflected posterolaterally. Through this exposure the entire posterior triangle is visible. For longer lesions the incision may be extended along the deltopectoral groove, if necessary.

we have had to extend the incision along the deltopectoral groove for exposure of the more distal plexus in long lesions.

Dissection

The procedure is performed under loupe magnification. The skin is elevated in the subplatysmal plane and retracted superolaterally. The lateral border of the sternocleidomastoid is identified, and the clavicular head is released from its origin. Along the lateral border of the sternocleidomastoid, the cervical plexus can be seen emerging and branching.³⁹ The branches should be neatly dissected⁴¹ (Fig. 53-9). Supraclavicular branches are often useful as graft material. As they are followed proximally while working toward the C3 and C4 roots, larger branches (the greater auricular, lesser occipital, and cervical cutaneous) can be identified. Early identification of the C4 root is helpful for proceeding with the exposure and identifying the brachial plexus roots proper (C5-T1). The omohyoid muscle divides the posterior triangle into a superior and inferior portion. The omohyoid should be divided at its tendinous midportion and reflected from the field. The dissection along the lateral border of the sternocleidomastoid can then continue. Beneath the omohyoid, a layer of adipose tissue and lymphatics is seen. This tissue directly overlies the injured plexus. It is best to mobilize this tissue as a flap by dissecting it from the lateral border of the sternocleidomastoid and then sweeping it off the clavicle in a medial-to-lateral direction. The flap, which is hinged posterolaterally, can be placed over the reconstructed plexus at the completion of the operation. The transverse cervical artery can be seen and is divided for further exposure. To prevent potential instability and the development of bone callous directly over the reconstruction, we do not divide the clavicle or remove a central segment as some do for the purpose of exposure. We generally proceed with exposing the

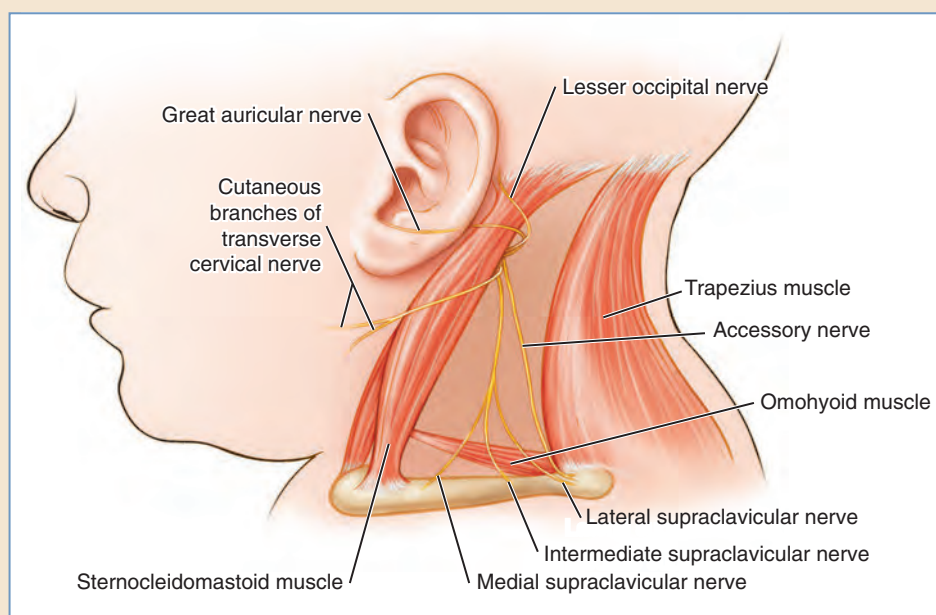


Fig. 53-9 The sternocleidomastoid, trapezius, and clavicle form the borders of the posterior triangle. The omohyoid divides the triangle into upper and lower portions. The accessory nerve travels obliquely. The branches of the cervical plexus are more superficial and include the lesser occipital, greater auricular, cervical cutaneous, and supraclavicular nerves. Supraclavicular branches further divide and can be useful as grafts, providing 3 to 4 cm of material.

distal plexus with the aid of an assistant, who adequately retracts the clavicle anteroinferiorly. If it is necessary to divide the clavicle, it can be restored as Borrero⁴¹ described, using a figure-of-eight suture passed through drill holes.

The plexus exits between the anterior and middle scalenes proximally. These muscles are often scarred and adhere to the neuroma on their respective sides. The anterior scalene is dissected from the neuroma, working toward the C5 foramen. Two important structures will be visible. The phrenic nerve travels inferiorly and should be carefully preserved, although any contribution from (and often to) the plexus can be divided. It is frequently scarred to the anterior surface of the neuroma.²⁶ Also directed inferiorly, but passing behind the neuroma, is the long thoracic nerve. The C5 root is dissected proximally all the way to its foramen. Dissection within the bony foramina at any level can cause bleeding from disruption of small vessels, primarily venous. This is generally controlled with time and pressure. The C6 and C7 roots are dissected next in a similar fashion. The absence of a particular root from its expected position could indicate that the root has been avulsed from the cord. The foramen should still be completely dissected to confirm this and perhaps to identify a proximal stump created by rupture rather than avulsion. The only irrefutable proof of avulsion is the identification of a dorsal root ganglion in the surgical field. The dorsal root ganglion can generally be identified by sight, appearing as a bulb of nerve tissue at the proximalmost end. If the dorsal root ganglion cannot be identified with certainty, a histologic examination is performed for a definitive diagnosis.

The upper and middle trunks, which are often difficult to separate, should be in view. Dissection of the lateral border of the plexus in a proximal-to-distal direction allows identification of the suprascapular nerve. Depending on the injury, this nerve may be seen branching from the posterior

division of the upper trunk proximally, distally, or within the zone of injury.⁴² Generally, the branching pattern of the plexus can be distorted by several factors: the length of the lesion, the presence of ruptures, the position of ruptures, and the extent that the neuroma has contracted. Contraction can be considerable, and this will alter expected branching positions. For example, the separation of the middle trunk into anterior and posterior divisions is normally retroclavicular, but this may occur more proximally in a lesion that has contracted.⁴¹ The lateral pectoral nerve, which arises from the anterior division of the upper trunk,⁴² is a useful landmark for distinguishing the anterior from posterior divisions of the upper trunk in a heavily scarred plexus. The dissection continues under the clavicle until all branches have been identified and the distal limits of the neuroma have given way to the normal branching network that will ultimately serve as graft targets.

The lower trunk, formed by C8 and T1, is dissected from distal to proximal. The subclavian artery can be found overlying the lower trunk. It should be dissected from the neuroma to be safely retracted for exposure of the C8 and T1 roots and foramina. The suprascapular artery can be intimately associated with the neuroma. It and the transverse cervical artery, which was more superficial and was already divided, are both branches of the thyrocervical trunk from the subclavian artery. Ligation and division of the suprascapular artery and vein are often necessary. Once these vessels have been appropriately addressed, the C8 and T1 roots are approached. The T1 root is smaller than the C8 root and lies posterior and inferior to it. Dissection of T1 is one of the most challenging portions of the dissection, because it lies on the parietal pleural surface. From an anterior approach, the T1 foramen is difficult to see. As the lower trunk is followed distally, all branches are located and preserved. The dissection again continues until the normal branch network distal to the lesion is identified. Unlike the upper and middle trunk components, which are often densely adherent, the lower trunk components are sometimes more easily dissected from the neuroma. The final portion of the exploration that must be completed is dissection of the posterior surface of the plexus/neuroma, which is often completed after resection of the damaged plexus.

NEUROLYSIS VERSUS RESECTION AND RECONSTRUCTION

The most common anatomic operative finding is neuroma-in-continuity, with variations in the length and the number and extent of involved roots. However, even though we see physical continuity, it is very difficult to know whether this is accompanied by regeneration of axons through the neuroma, that is, physiologic continuity. Although some authors advocate intraoperative neurophysiologic testing, we have found that functionally relevant quantification of physiologic continuity is not possible with current technologic measures. This may be why neurolysis of the plexus has not proved to be a useful means of managing obstetrical brachial plexus lesions. As Borrero⁴¹ indicated, most surgeons with experience agree that results after neurolysis alone are discouraging. Earlier in our practice, we performed neurolysis when a conducted stimulus across the neuroma produced an appropriate distal muscle contraction. We reported the results in 16 patients who underwent neurolysis, comparing preoperative active motion with that seen at the 12-month follow-up.⁴³ We observed improvement in the group of patients with upper plexus lesions, but it was ineffective in patients with total plexus palsy. We abandoned neurolysis in patients with total plexus palsy in favor of neuroma resection and grafting. Eventually, we abandoned neurolysis in patients with primarily upper plexus lesions when we recognized that infants who were treated with resection appeared to regain baseline function rapidly after resection. In 1998 we reported preoperative and postoperative active motion over time in 26 patients treated with a neuroma resection.⁴⁴ The results showed an initial decrease in function compared to the baseline. However, by 3 months no difference was noted in active motion from baseline, and by 6 months patients had either reached or surpassed the baseline. The difference could not be explained on the basis of regeneration across grafts at these early time points. Furthermore, the early follow-

up results (at 3, 6, and 12 months) were no worse for those who had undergone resection than for patients who had undergone neurolysis. Based on the observation that neuroma resection is not detrimental, and the growing body of evidence from Birch,¹² Kawabata et al,⁴⁵ Gilbert et al,^{46,47} and Aydin et al⁴⁸ that grafting offers the best opportunity for maximal functional recovery, we have strongly advocated neuroma resection and grafting as the primary management of the neuroma in obstetrical palsy.

In 2009 we published our long-term results of neurolysis versus neuroma resection and grafting with a minimum 4-year follow-up.⁴⁹ After neurolysis, patients with Erb palsy and total palsy demonstrated no improvement in function (Fig. 53-10, *A* and *C*). By contrast, patients with Erb

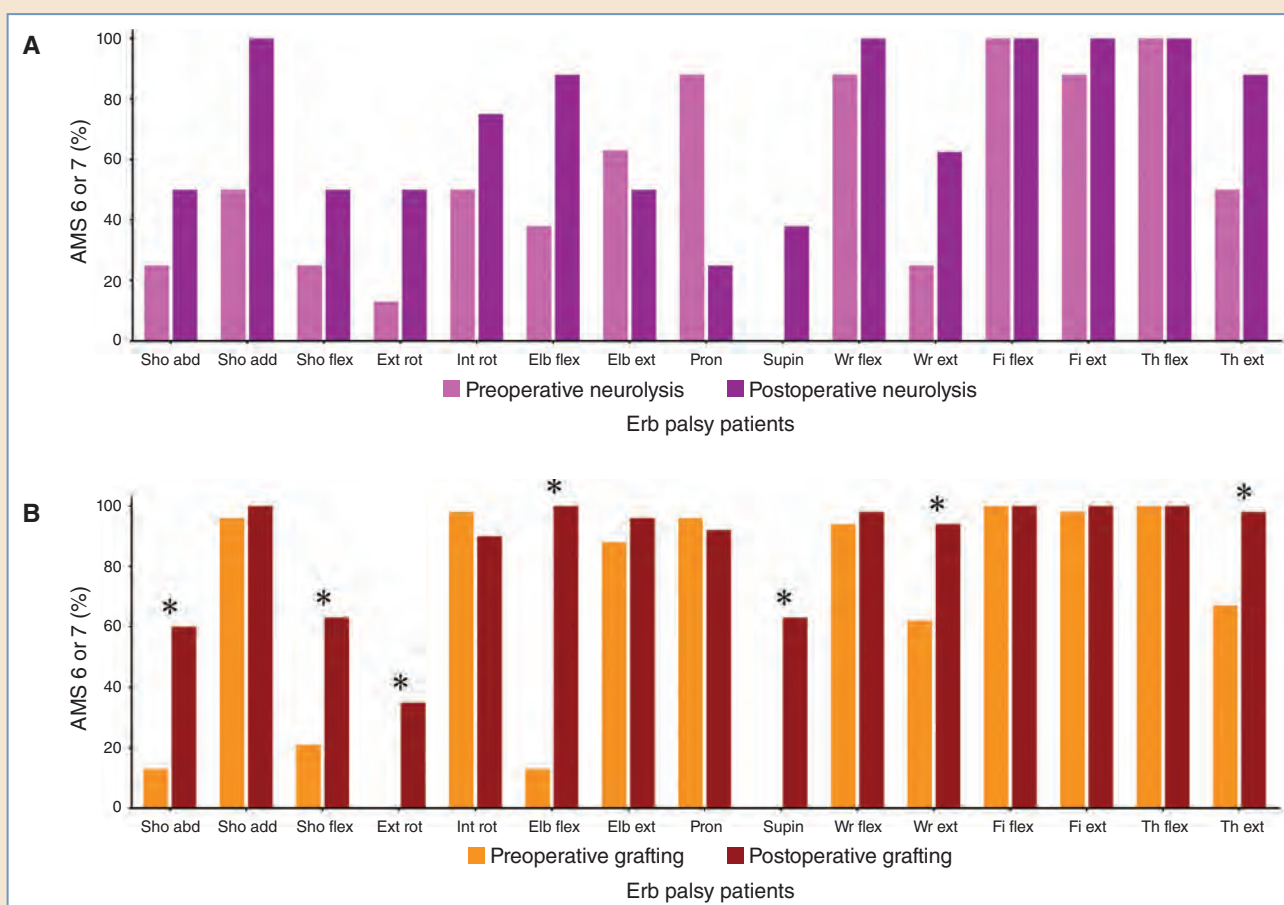


Fig. 53-10 **A**, Preoperative proportions of patients obtaining an Active Movement Scale (AMS) score of 6 or 7 for 15 movements of the upper extremity were compared with final proportions at least 4 years after a neurolysis for Erb palsy. (* $p < 0.05$ [McNemar exact test].) **B**, Preoperative proportions of patients obtaining an Active Movement Scale score of 6 or 7 for 15 movements of the upper extremity were compared with final proportions at least 4 years after neuroma resection and interpositional nerve grafting for Erb Palsy. (* $p < 0.05$ [McNemar exact test].)

palsy and total palsy undergoing neuroma resection and grafting had significant improvements in most of the affected upper extremity motions, as measured using the Active Movement Scale⁴⁹ (Fig. 53-10, *B* and *D*). This study provided strong evidence that neurolysis as a complete surgical treatment for obstetrical brachial plexus palsy should be abandoned in favor of neuroma resection and grafting. Neurolysis can be useful in selected patients in whom a normal fascicular pattern is demonstrated within surrounding scar. This can occur in upper trunk or upper and middle trunk injuries that result in scarring around otherwise uninjured C8 and T1 roots. However, unless a distinct, normal fascicular pattern is evident, and direct nerve stimulation produces appropriate muscle contractions, we resect all lesions and proceed with reconstruction using nerve grafts.

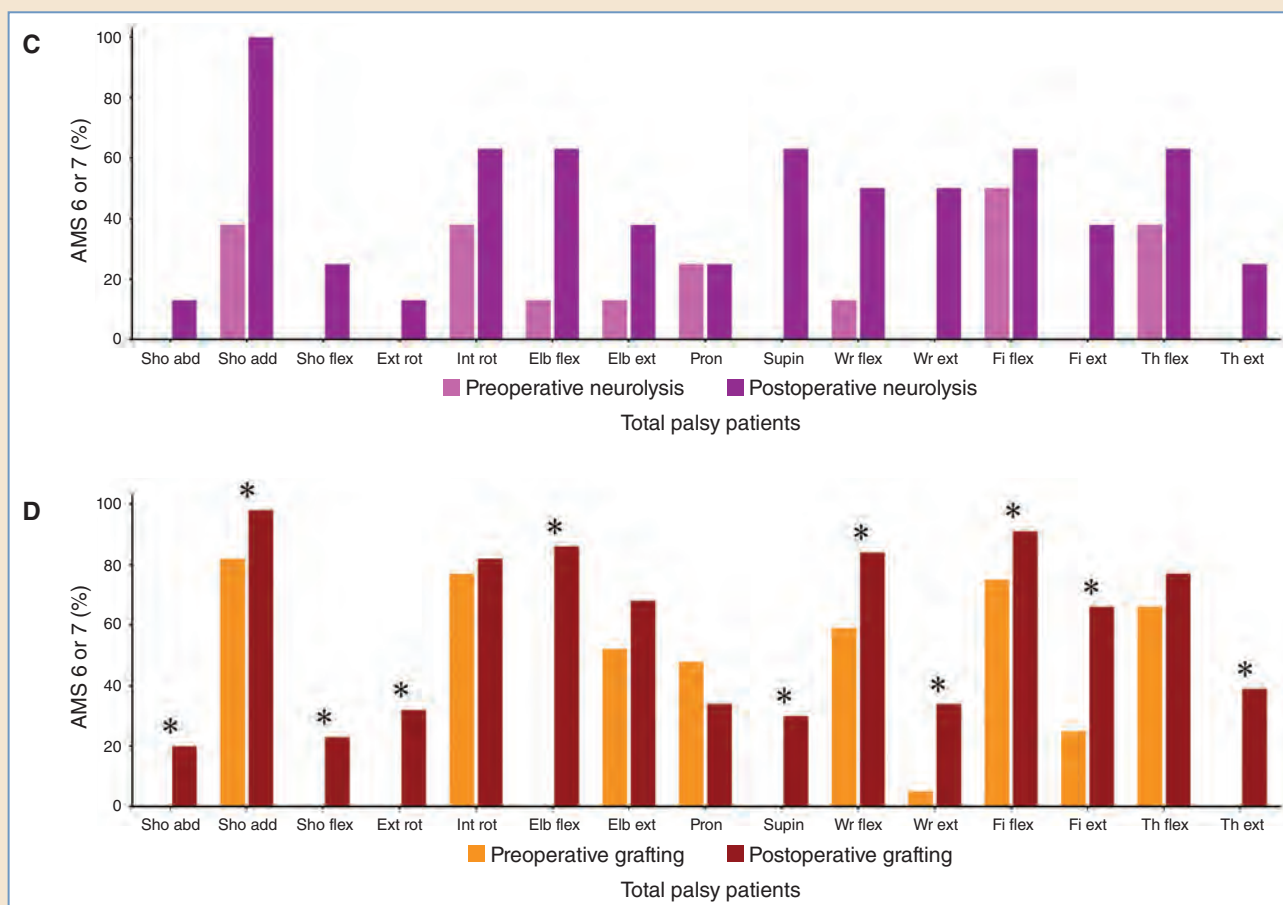


Fig. 53-10, cont'd **C**, Preoperative proportions of patients obtaining an Active Movement Scale score of 6 or 7 for 15 movements of the upper extremity were compared with final proportions at least 4 years after neurolysis for total palsy. (* $p < 0.05$ [McNemar exact test].) **D**, Preoperative proportions of patients obtaining an Active Movement Scale score of 6 or 7 for 15 movements of the upper extremity were compared with final proportions at least 4 years after neuroma resection and interpositional nerve grafting for total palsy. (* $p < 0.05$ [McNemar exact test].) (*Elb ext*, Elbow extension; *Elb flex*, elbow flexion; *Ext rot*, external rotation; *Fi ext*, finger extension; *Fi flex*, finger flexion; *Int rot*, internal rotation; *Pron*, pronation; *Sho abd*, shoulder abduction; *Sho add*, shoulder adduction; *Sho flex*, shoulder flexion; *Supin*, supination; *Th ext*, thumb extension; *Th flex*, thumb flexion; *Wr ext*, wrist extension; *Wr flex*, wrist flexion.)

Evaluation of the Proximal Roots

Exploration and dissection of the plexus ideally should reveal a central traumatized area, represented by a neuroma, with normal structures proximally and distally. The distal dissection should always continue until grossly normal, identifiable branches are exposed. The length of the neuromatous lesion determines whether this occurs at the level of divisions, cords, or terminal branches. We typically transect the neuroma through its midportion. Each of the distal branches is transected at a level that appears grossly normal, and the distal half of the neuroma is removed. These carefully identified ends are the targets for sural nerve grafts. Depending on its branch point location, the suprascapular nerve is usually an independent distal target. The condition, quality, and axonal topography of the distal ends are demonstrable through histologic frozen section using toluidine blue dye. This is always performed in our practice.

Proximally, a major dilemma can occur. An empty foramen, or histologic evidence of a dorsal root ganglion outside the foramen with otherwise apparent continuity, irrefutably indicates root avulsion from the spinal cord. With each avulsion, fewer proximal source stumps are available to provide axons to the distal targets. When continuity from within a particular foramen along the neuroma is apparent, how do surgeons definitively know whether the proximal stump will be a suitable source? How can we determine whether an intraforaminal avulsion has occurred, if it is not visible? This is the most vexing problem in the operation, because grafts from a stump that appears normal (grossly and even histologically) will provide no axons whatsoever if it has been avulsed from the spinal cord. The only way to know with certainty whether an intraforaminal avulsion has occurred is through a laminectomy. However, because this is generally not considered safe for infants, we must weigh all available evidence to reach a most probable conclusion. Box 53-1 presents an outline for the available evidence.⁴¹

Some of this evidence warrants further discussion. Although using MRI to evaluate root status has been advocated in recent reports,^{50,51} technical difficulty, reliance on exquisite interpretive skills, and the need for further statistical validation have limited the widespread application of MRI for determining root avulsion. We continue to investigate all infants preoperatively with

Box 53-1 Steps for Determining an Intraforaminal Avulsion

1. Preoperative data
 - a. A physical examination
 - i. The Hospital for Sick Children's Active Movement Scale (in our practice)
 - ii. The presence of Horner sign
 - b. Radiographic data: CT myelography (or MRI in some centers)
2. The muscle response to direct stimulation of the root. A normal-appearing nerve will not conduct distally if complete avulsion has occurred.
3. The operative findings: the gross appearance of the stump, tactile sensation during transection
4. A histologic examination of the proximal stump
5. Intraoperative electrophysiologic testing

CT myelography. A CT myelogram provides useful information depending on the presence of a pseudomeningocele and the ability to see rootlets traversing it. Chow et al⁵² studied 281 roots in 58 patients undergoing exploration and reconstruction. Comparing radiographic interpretation to findings during surgery, they determined the sensitivity, specificity, positive predictive value, and the likelihood ratio for these two possible indicators of root avulsion. Of 38 root avulsions, 24 were associated with pseudomeningocele, for a low sensitivity (0.63) but a relatively high specificity (0.85). For pseudomeningocele with absent rootlets, the sensitivity was even lower, but the specificity was near perfect (0.98), and the likelihood ratio was 18.5. A low sensitivity indicates that avulsion may be present without either of these findings. With high specificity, however, the proportion of false-positives, or normal roots incorrectly identified on a CT myelogram, decreases. Therefore a positive finding (a pseudomeningocele with absent rootlets) is more suggestive of root avulsion at a particular level. In other words, when rootlets cannot be seen in association with a pseudomeningocele, a false-positive (a normal root) is highly unlikely; it is 18.5 times more probable that the involved root is avulsed (Fig. 53-11). Because the sensitivity is low, further evidence is needed to rule out avulsion in the absence of these statistically specific indicators.

Recent evidence has suggested that intraoperative electrophysiologic testing is unreliable as a predictor of root avulsion. In a randomized, prospective, double-blind trial, Dupere et al⁵³ showed no correlation between the findings of an expert neurophysiologist evaluating root avulsions with somatosensory evoked potentials and surgical findings.

Histologic analysis of the root stumps before grafting has proved useful. When a neuroma is transected proximally, the site must be proximal to the injury to allow axonal growth from the stumps to coapted grafts. Visual inspection of the cut stump end is insufficient to determine the status of the proximal stump neural architecture. Redett et al⁵⁴ examined the proximal stumps after transection guided by visual inspection. The authors demonstrated that frozen-section analysis of the root stumps led to a measurable improvement in the histologic grade of the stumps after re-resection in appropriate candidates. Frozen-section analysis prompted re-resection to reach a proximal level suitable for grafting in at least one stump in 25% of patients overall.



Fig. 53-11 A preoperative myelogram, axially at the level of C8, shows a pseudomeningocele at C7-T1 (arrow). Neither dorsal nor ventral C8 rootlets are seen traversing the pseudomeningocele. This finding is a very specific indicator of root avulsion (specificity 0.98), with a likelihood ratio of 18.5 for avulsion.

Grafting

The neuroma has been resected, the targets have been identified, and their proximal and distal ends histologically deemed satisfactory. The usable (decidedly nonavulsed) proximal stumps have been selected. A gap has been created. Multiple strands of sural nerve graft will span this gap. If the supraclavicular branches of the cervical plexus have been carefully dissected, these may also serve as graft material.

Sural Nerve Graft Harvest

We elect to harvest bilateral sural nerves at the first stage of the operation (that is, before exploration of the plexus). We perform sural nerve harvest using endoscopic assistance. Capek et al⁵⁵ provided a complete description. The infant is prone for this portion of the procedure and is then placed in a supine position for exploration and reconstruction of the plexus. In this way, exposure and access are optimal, and only one change in position is required. An endoscopic-assisted harvest is performed through three 2 cm transverse incisions⁵⁵ (Fig. 53-12). The sural nerve can be harvested from its most proximal origin from the tibial nerve to the most distal branches supplying the lateral foot. In an 8 to 10 kg infant, an average of 13 to 15 cm can be harvested per leg. The harvested nerves are placed in moist, sealed, sterile containers that are refrigerated until they are needed.

Graft Reconstruction Planning

A reconstruction plan is formulated and individualized to the unique anatomic findings of each patient. Variations are seen in each of the following:

- The positions of targets distally along the branching plexus
- The number of available proximal source stumps
- The presence and extent of injury to the lower plexus
- The length of the neuroma and therefore the length of needed graft

If a target is reconstructed using grafts from its developmentally appropriate source root, then the reconstruction is anatomic grafting for that target. If at all possible, this is the first choice for reinnervation of target nerves. If, however, the appropriate source root is avulsed, then associated

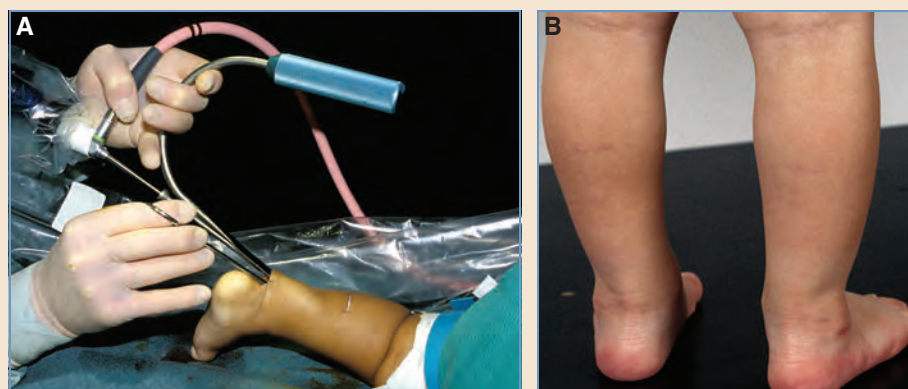


Fig. 53-12 An endoscopic sural nerve harvest, as described by Capek et al. **A**, A series of three 2 cm transverse incisions are made, centered along the midline of the calf. The nerve is harvested distally along the lateral aspect of the foot and proximally to its origin from the tibial nerve, where additional length can be obtained by intrafascicular dissection. **B**, The resultant scars from harvest in this manner are generally very acceptable and prevent a long "stocking seam" incision.

targets must receive innervation from an alternate source, a procedure known as *neurotization*. This can be accomplished with remaining roots (plexoplexal or intraplexal neurotization) or from nerve donors outside the plexus (extraplexal neurotization).

The distances to every target are measured from each of the available roots. The length of available graft material (sural nerve and cervical plexus) likewise is measured. The division and allocation of graft material is prioritized. Any of the above-noted variations can present limitations, and reconstruction requires consideration of the greatest functional needs. Most investigators agree that the use of the upper extremity depends on a functioning hand. Therefore prioritization of functional motion should first focus on reinnervation of the hand.^{28,41} This is addressed using multistrand grafting to lower trunk targets. After the hand, priority is directed to elbow flexion and then to shoulder motion.

Grafts are placed into position in reversed orientation to prevent progressing axons from prematurely exiting the graft through side branches of the original sural nerve. Graft placement is performed under an operating microscope. In our practice, nerves are coapted using commercially available fibrin glue, with no sutures. Some surgeons prefer to use suture material only, and others use a single suture reinforced with fibrin glue for end-to-end coaptation.

Fig. 53-13 depicts the working plan and the final diagram for a patient with upper plexus palsy with avulsion of C8. On average, the distance from most sources to the respective targets is 2.5 to 4.5 cm, as in this case. T1 had a distinct and normal-appearing fascicular pattern and was

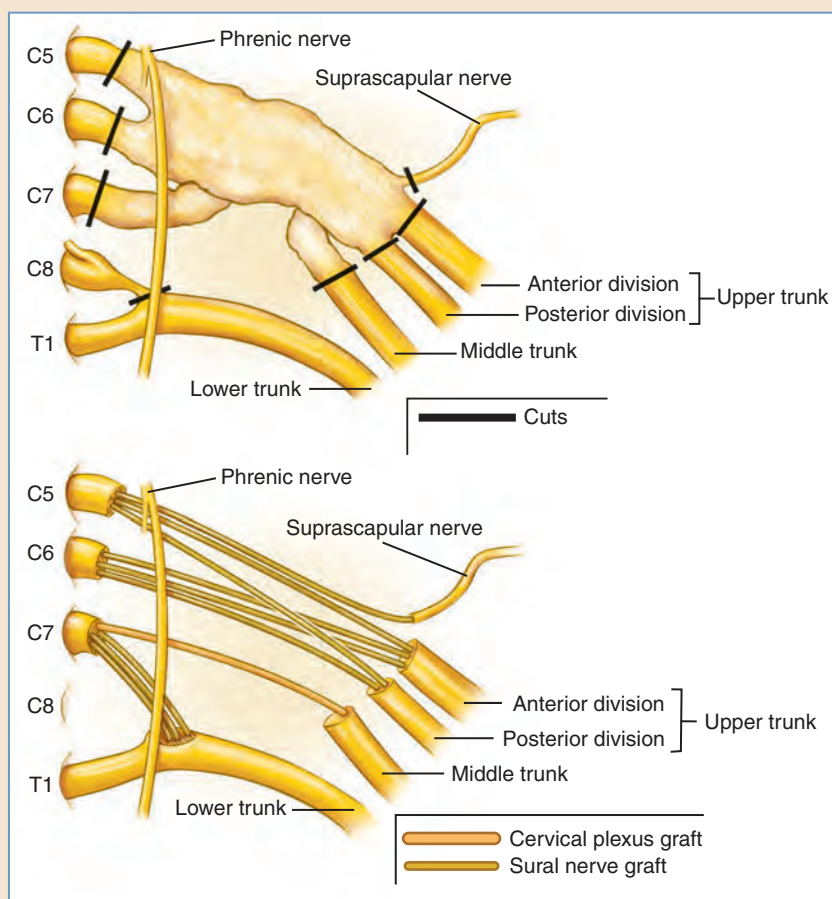


Fig. 53-13 This patient presented with an upper plexus palsy and was found to have an avulsion of C8, demonstrated unequivocally by the presence of a dorsal root ganglion. C5-7 was involved in a dense neuroma, and T1 had a normal fascicular pattern with minimal involvement in the neuroma. Anatomic grafting was performed to the suprascapular nerve, anterior and posterior divisions of the upper trunk, and the middle trunk. Intraplexal neurotization to the lower trunk was performed from C7. T1 was treated with a neurolysis. The phrenic nerve is shown traversing longitudinally over the plexus.

treated with a neurolysis. The remaining distal targets, including the suprascapular nerve, were reconstructed with grafts from the three intact stumps. For most targets, grafting was anatomic. Grafts to targets that had received C8 innervation (avulsed) are considered intraplexal neurotizations, because they were grafted from other available stumps.

Fig. 53-14 presents a patient with avulsion of C7, C8, and T1. In this case a longer gap was created after a neuroma resection, making strategic graft allocation even more important. The lower plexus targets were given first priority (highest priority to hand function) to receive grafts from the available C5 and C6 stumps. The lower trunk therefore was reconstructed using an intraplexal (nonanatomic) neurotization. The upper trunk targets were selected next to favor elbow flexion and shoulder function.

This case also demonstrates the use of extraplexal neurotization. The suprascapular nerve was reconstructed from the spinal accessory nerve (cranial nerve XI). This is the most commonly

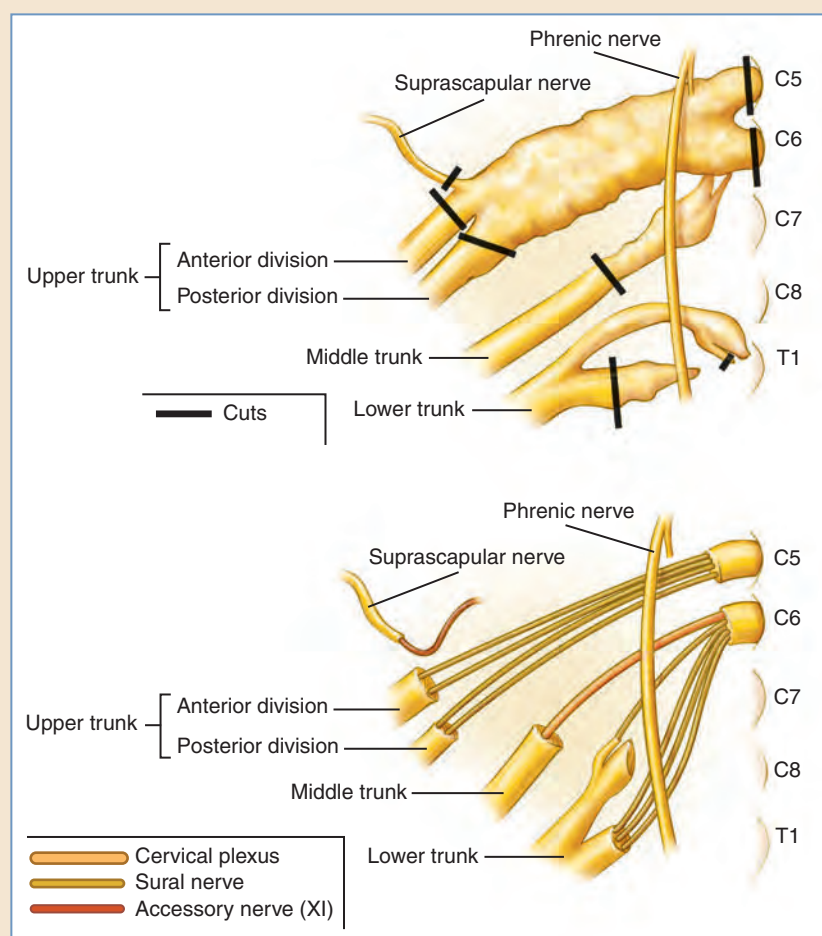


Fig. 53-14 This patient presented with a total plexus palsy and was found to have avulsions of C7, C8, and T1. A long lesion was present. This was a difficult situation because of the paucity of stumps available for grafting and the long lengths of graft that were required. The lower trunk targets were given first priority to provide hand function and were grafted from the C6 stump, as was the middle trunk. The anterior and posterior divisions of the upper trunk were grafted from C5. Extraplexal neurotization of the suprascapular nerve from the accessory nerve was a useful maneuver to conserve available graft material.

performed extraplexal neurotization, according to most authors. Cranial nerve XI is dissected distally beneath the trapezius muscle to its major branch point. The dissection is performed through the existing neck exposure. The longitudinal branch supplying the thoracic portion of the trapezius is divided distally and transposed into the field, where it consistently reaches the cut end of the suprascapular nerve free of tension. Performed in this manner, the function of the cervical/upper thoracic portion of the trapezius is not sacrificed. This muscle inserts on the clavicle, acromion, and scapular spine. The suprascapular nerve provides external rotation at the shoulder through the actions of the infraspinatus and supraspinatus muscles. Tse et al⁵⁶ have shown that neurotization using the spinal accessory nerve produces external rotation at the shoulder equal to that obtained by anatomic reconstruction using sural nerve grafts from C5. Other extraplexal neurotizations include the intercostal nerves (typically two to three donors are used) and the contralateral C7 root. Before performing an extraplexal neurotization, surgeons should consider the expected potential gain in function and the donor deficit.

Postoperative Care

Postoperatively, a simple dressing is applied over the incision, and the child is fitted with a Velpeau sling made with a single length of 2-inch stockinette material (Fig. 53-15). Properly fitted and maintained, this soft, inexpensive bandage effectively maintains the arm in full internal rotation and shoulder adduction with the elbow flexed to 90 degrees.⁵⁷ We have used this technique and have appreciated its efficacy and advantages over plaster immobilization in more than 250 primary infant plexus reconstructions. Infants tolerate it extremely well, and parents and nurses can easily maintain and adjust it. A Velpeau sling is worn continuously for 3 weeks.

Complications

Both major and minor complications can occur with brachial plexus reconstruction. In our recent review of 173 brachial plexus reconstructions for obstetrical palsy, the total combined rate of major and minor complications was 33.5% (58 patients).⁵⁸ The major complication rate was 18.5%, and the minor complication rate was 15%. The incidence of complications did not appear to be



Fig. 53-15 A light dressing is placed on the incision, and a soft stockinette Velpeau-style garment is fashioned to keep the arm securely adducted, with the elbow bent to 90 degrees. The sling is worn continuously for 3 weeks. It is not necessary to employ a plaster shoulder girdle casting with this technique.

influenced statistically by the age of the patient (ranging from 3 months to 1 year). No deaths occurred. Major complications included wound infection (2.3%), seroma (1.2%), and respiratory complications of postoperative hemidiaphragm paralysis (6.4%), atelectasis (2.9%), pulmonary edema (1.7%), pleural effusion (1.7%), pneumothorax (1.7%), and chylothorax (0.6%). Accidental extubation occurred in five patients in the early part of the series. With the addition of a clear plastic head draping and suture fixation of the nasotracheal tube to the membranous septum, this has not occurred in the subsequent 100 patients. Pneumothorax was managed intraoperatively in two of the three affected patients by pleural repair and reinforcement. This was assisted by placement of a rubber catheter to apply negative pressure; repair was confirmed by a submerged Valsalva maneuver. In the third patient, a pneumothorax was identified by radiographic examination in the recovery area and was treated with aspiration.

Long-term complications associated with sural nerve harvest are minimal. In a controlled study of patients undergoing sural nerve harvest for brachial plexus reconstruction, 86% demonstrated a measurable sensory deficit in the lateral aspect of the foot at a minimum of 5 years postoperatively.⁵⁹ However, no patients reported sensory problems, and no functional implications are related to this sensory deficit.

NERVE TRANSFERS

Extraplexal and distal nerve transfers may be indicated for patients with a delayed presentation in a delayed fashion, multiple cervical root avulsions, or insufficient graft material for completing the desired reconstruction. The most commonly used extraplexal transfer is the spinal accessory to suprascapular nerve transfer to restore external rotation, as described previously.⁶⁰ Successful neurotization of the myocutaneous nerve to restore elbow flexion using the thoracic intercostal nerves or the medial pectoral nerve has been described.⁶¹⁻⁶³ Other extraplexal donor nerves include the phrenic nerve and, in the rare situation of a five-root (total) avulsion, the contralateral C7 nerve root.^{64,65}

More recently, the distal nerve transfers described for traumatic brachial plexus reconstruction in adults have been applied to Erb palsy. Al-Qattan⁶⁶ was the first to publish his results on the use of the ulnar-to-biceps nerve (Oberlin et al⁶⁷) transfer in patients with poor elbow flexion presenting later than 12 months of age.

Other surgeons have published small case series in which the ulnar or median nerve was used to neurotize the biceps and/or brachialis nerve for primary or late elbow reconstruction in patients up to 24 months of age.^{66,68-70} For restoration of shoulder motion, the triceps-to-axillary nerve transfer and the posterior approach to the spinal accessory-to-suprascapular nerve transfer have been described.⁷¹⁻⁷⁴

For many surgeons, distal nerve transfers are more appealing than proximal neuroma resection and grafting because of their greater technical ease, reduced operative time, and earlier reinnervation of the target muscle. Although distal nerve transfers play a role in selected cases of obstetrical brachial plexus palsy, caution must be exercised in adopting this approach for all patients. One of the major disadvantages of distal motor nerve transfers is the lack of sensory reconstruction. Furthermore, in upper and middle trunk injuries, selective reconstruction of elbow flexion and shoulder abduction may not enhance or restore other affected motions such as pronosupination. Without proximal exploration, the extent of the lesion may be underappreciated, and the opportunity for neurolysis of uninjured roots (for example, C8 and T1) from the scar generated by an upper and/or middle trunk injury is lost. Finally, no evidence has been reported to support the long-term functional outcomes of distal nerve transfers in Erb palsy patients. Future studies that rigorously evaluate the role of nerve transfers in primary nerve reconstruction in these patients will help to determine the indications for this approach.

CONCLUSION

Primary microsurgery for obstetrical brachial plexus lesions is a new area of surgical expertise. It offers the possibility of improved functional ability in carefully selected patients who would otherwise have lifelong impairment and secondary skeletal deformities. One major challenge in this area of peripheral nerve surgery is the selection of patients most likely to benefit from surgical intervention. Key factors for developing selection criteria and resolving other issues (such as identifying root avulsions) include consistency, accuracy, and careful reporting of natural history and outcome data. In particular, we strongly emphasize that a statistically sound method of assessment must be consistently applied from the time of presentation through long-term follow-up. Advancement, to date, has resulted from the application of evidence-based recommendations from well-designed studies. As the field of obstetrical brachial plexopathy management continues to evolve, questions will continue to be answered using such scientific methodology.

KEY POINTS

- Most patients with an obstetrical brachial plexus lesion do not require surgery.
- Determining the need for surgery is the most difficult aspect of treatment and relies on a thorough understanding of the natural history of such injuries and careful application of selection criteria.
- Selection criteria must be based on validated, objective test instruments. Such instruments should be subjected to rigid statistical analysis for the purpose of formulating a reliable prognosis.
- A detailed obstetrical history and physical examination provide useful data for determining the spinal root injury level and the proximal-to-distal extent of an injury.
- Appropriate retraction of the clavicle generally obviates the need to divide it for distal exposure.
- Neurolysis may be appropriate only for conditions in which a distinct fascicular pattern is seen within the surrounding scar; otherwise, statistical evidence indicates that neuroma resection and grafting offer the best opportunity for maximal functional recovery.
- Proximal stumps and distal targets should be histologically evaluated before grafting to determine suitability for grafting.
- Neurotization techniques vary and provide useful means of neural input to selected targets.
- A Velpeau sling provides efficient and tolerable immobilization postoperatively for a recommended duration of 3 weeks.

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54

Volkmann Ischemic Contracture

Ronald M. Zuker • Joseph Upton III



Volkmann ischemic contracture is a devastating injury. In children it takes two forms. The first, which is known as *neonatal Volkmann contracture*, is present at birth and is usually the result of intrauterine emboli, compression, or a combination of both. Various degrees of skin loss and muscle loss are present, requiring complex wound management and reconstruction. Secondary effects on neurologic function and skeletal growth can be devastating.

The second form, *childhood Volkmann ischemic contracture*, usually results from limb trauma, most commonly a supracondylar fracture. Forearm ischemia results, which leads to muscle necrosis and subsequent fibrosis and contracture formation. In this chapter, both forms are discussed. We describe the clinical picture, the pathophysiologic processes involved, and the management of these complex conditions, which often includes muscle replacement using microvascular techniques.

Neonatal Volkmann Ischemic Contracture

DEFINITION OF THE PROBLEM

Neonatal compression injuries and compartment syndromes causing secondary ischemic contractures are rare and almost universally misdiagnosed.¹⁻⁶ The spectrum of anatomic severity is wide and extends from dorsal skin bullae to circumferential full-thickness skin loss and ischemia of the dorsal and volar compartments in the proximal two thirds of the forearm.

This condition is not listed as a cause of compartment syndrome in the most recent edition of commonly used texts of hand surgery.⁷ We have included this condition as a compartment

syndrome because of the characteristic prenatal history and physical examination of the newborn. The pathophysiology is the same as that of compartment syndrome after arterial compromise associated with the supracondylar fracture in a child or a crushing injury to the forearm seen after trauma.

The literature on this condition consists primarily of case reports, small series, and case reports with a summary of the literature to date.⁸⁻²³ Based on the numerous reports, we advocate that this condition no longer be considered as rare, but rather one that is not easily diagnosed. This chapter is based on 49 cases I (J.U.) have seen over a 40-year period in a large children's hospital, which is a tertiary referral center for the New England states. We conclude that this condition, especially the milder forms, is often either misdiagnosed or not recognized early in life.

Differential Diagnosis

The differential diagnosis of skin maceration leading to ulceration in newborns can be very confusing. There are no concise or comprehensive discussions in the surgical or pediatric literature, and the underlying causes of these problems are constantly overlooked. Arterial compromise is usually associated with failed arterial lines, repeated arterial or venous punctures, or trauma with kinking and thrombosis of vessels. More diffuse skin eruptions, bullae and small vesicle formation, and skin macerations and lesions are seen with various types of cutis aplasia, congenital epidermolysis bullosa, and other conditions primarily confined to the integument. Many conditions can be differentiated from neonatal Volkmann ischemia or compartment syndrome (Table 54-1).

Associated involvement of other portions of the body can yield these diagnoses. Distal arterial insufficiency resulting in amputation is often described as a vascular catastrophe and is usually associated with low-flow states in infants who are in shock or on extracorporeal membrane oxygenation machines. Extravasation of irritating intravenous infusions can cause significant local edema and potential tissue loss. In this condition, history and physical examination findings are very specific, and the extravasations are usually well documented.

Table 54-1 Conditions Differentiated From Neonatal Volkmann Ischemia

Condition	Symptomatology	Methods of Diagnosis
Neonatal gangrene	Associated with cool limb and decreased pulses, predisposing catheterization, indwelling lines, or hematologic problems (polycythemia)	Angiography, CTA, Doppler ultrasonography
Necrotizing fasciitis	Fever, warmth, sepsis	Blood cultures, biopsy, surgery
Cutis aplasia congenital	Associated scalp necrosis, digital amputations, and hypoplasia; healed and not healed ulcers	Biopsy, clinical examination
Amniotic band syndrome (constriction ring syndrome)	Associated bands, congenital amputations, acrosyndactyly	Clinical examination
Subcutaneous fat necrosis	Associated red to blue plaques, nodules on the trunk and limbs, not present at birth	Skin biopsy
Epidermolysis bullosa	Very fragile skin with bullae	Skin biopsy, including electron microscopy studies and immunofluorescent mapping

Background Information

The name *Volkmann ischemic contracture* of the forearm may be misleading, because many newborn infants with this condition have not developed secondary contractures.^{24,25} However, the eponym is used in this chapter, because it is easily recognized and the pathophysiology is similar. Similar cases are published in the literature under the names *neonatal Volkmann ischemia*, *neonatal compartment syndrome*, *extremity gangrene in utero*, *neonatal forearm compartment syndrome*, *acute ischemia of the newborn*, *neonatal arterial occlusion*, and *neonatal compression syndrome*. Many of these cases are in the dermatologic literature listed under *cutis aplasia congenital*, which is a very specific syndrome with scalp and usually hand findings (see Table 54-1).

Genetics

Volkmann ischemic contraction has no known genetic association or predisposition. It is classified as an in utero disruption. The incidence seems to be much higher with multiple births and with amniotic fluid problems such as oligohydramnios. Isolated gestational conditions have been reported, but none are consistently found in the large cohort of patients.¹¹

CLASSIFICATION

No known classification system exists for this condition other than what we reported in the first edition of this book. Few publications have been devoted to this subject; most reports describe the initial presentation and diagnosis, and even fewer discuss specific treatment and long-term outcomes.¹⁻²³ We have separated our cases into five clinical groups (types) that are distinguished from one another by the extent of the compression of the soft tissue structures. The mildest form, type I, probably occurs more frequently but is not recognized because of the limited amount of skin pathology that is present at birth. We have not seen many of these cases clinically in our tertiary referral centers, where infants are referred for large, nonhealing neonatal ulcers. Box 54-1 describes the classification types, and Table 54-2 summarizes our classification system.

Box 54-1 Neonatal Volkmann Ischemic Contracture Types

- Type I: Partial-thickness skin bullae are present, with involvement of the dorsal extensor compartment. Arterial, neural, and skeletal anatomy is normal.
- Type II: Full-thickness skin ulcers or eschars are present, affecting both the dorsal and volar compartments. Involvement of the volar compartment is very subtle on physical examination. Contractures develop in the dorsal and volar compartments, and all patients show long-term changes in growth patterns.
- Type III: Both the volar flexor and dorsal extensor compartments are involved, and full-thickness loss in part of the dorsal compartment, with exposed periosteum, can occur. The nerves and arteries within the volar compartments are intact. Nerves can undergo compression neuropathies.
- Type IV: Much larger skin deficits signify severe compartment involvement and loss of one major artery, usually an ulnar artery, and possibly the ulnar nerve. Skin bullae and artery loss may be circumferential. Bone is exposed, and signs of digital ischemia are usually present.
- Type V: The most severe group is characterized by more extensive loss of all soft tissue and by osteomyelitis. Skin loss is circumferential. A partial or complete loss of one or more digits occurs consistently.

Table 54-2 Classification System for Neonatal Volkmann Ischemic Contracture

Type	Skin	Muscle Compartments	Nerve	Arterial (pulses)	Skeletal Early Findings	Skeletal Growth
I	Bullae only	None to one involved (usually dorsal)	Median and ulnar nerves both intact	Radial and ulnar pulses intact	Normal	Normal growth
II	Bullae; localized, full-thickness loss	Both	Both intact	Both intact	Normal	Moderate shortening (<25%)
III	Large, full-thickness loss	Both	Both intact	Both intact	Periosteum exposed	Moderate to severe (25% to 50%)
IV	Large to circumferential tissue loss	Both	One out (usually ulnar)	Both intact to one out	Cortex exposed $\pm$ digital loss	Severe (>50%)
V	Large to circumferential tissue loss	Both	One out (usually ulnar)	Both intact to one out	Cortex exposed; osteomyelitis; $\pm$ digital loss(es)	Severe (>50%)

PATHOPHYSIOLOGY

The primary pathologic process resulting in a contracture is an increase in pressure within a muscle compartment, producing irreversible ischemia, which leads to contracture and functional loss. Arterial ischemia may be the precipitating cause. We think this is true in 20% of our patients, who also present with cerebral embolic ischemia (neonatal stroke). Without citing the events that occur at a molecular level, we think the inciting trauma is pressure on the neonatal forearm, resulting in an increased compartment volume that leads to the familiar cascade of increased compartmental pressure, venous stasis, ischemia, muscle loss, fibrosis, and contracture. External pressure on these limbs that is greater than the normal systolic pressure will directly cause muscle necrosis. In limbs with a significant amount of myonecrosis, with or without skin ulceration, significant contractures can develop within a very short period (see Figs. 54-3, 54-4, and 54-8).

ANATOMY

The muscle and skin anatomy in these neonatal limbs is different from that seen in patients with classic, established Volkmann contracture caused by arterial occlusion. As the compartment pressure increases, the central portion of the muscle is more ischemic, and perfusion at the edges is retained. Muscle necrosis and degeneration occur more frequently in the central portion, and an elliptical band of scar forms parallel to the long axis of the forearm. The muscles most affected in the classic contracture are the deep and superficial flexors and the pronator teres. In contrast, these limbs with neonatal compartment syndrome have soft tissue damage and secondary contracture on the dorsoulnar aspect. It is localized to the direct site of compression and forms a dense block of fibrous tissue.

FUNCTIONAL CONSIDERATIONS

Because all tissues within the forearm and hand may be affected, the functional losses are wide ranging. In most established cases of pediatric and adult Volkmann contractures, the major sensory nerves are usually spared, and sensation in the hand is normal in all but the type IV and type V categories. The ulnar nerve is affected more commonly than the radial nerve. Because both the dorsal and volar muscle compartments are involved in all but the type I limbs, flexion and extension contractures are present. Forearm pronation and supination are usually limited in most and absent in severe types. In patients followed through childhood and adolescence, all but the type I limbs demonstrate growth abnormalities not only at the direct site of forearm compression but also at the distal growth plates of the radius and ulna (see Figs. 54-2, 54-6, and 54-8).

PREOPERATIVE ASSESSMENT

Maternal History

In our experience, no predisposing associated diseases, medical conditions, medications, or diagnostic procedures had complicated the early pregnancy. However, multiple births, mostly twins, was a common factor in more than a third of the cases. Labor was difficult and prolonged in all cases, and the overwhelming majority of infants were delivered by cesarean section. One infant, delivered vaginally by a midwife after a 40-hour labor, had loss of both radial and ulnar arteries resulting in the loss of all digits and both thumbs. Other factors include maternal varicella infection, gestation and insulin-dependent diabetes, prematurity, and coagulation abnormalities.

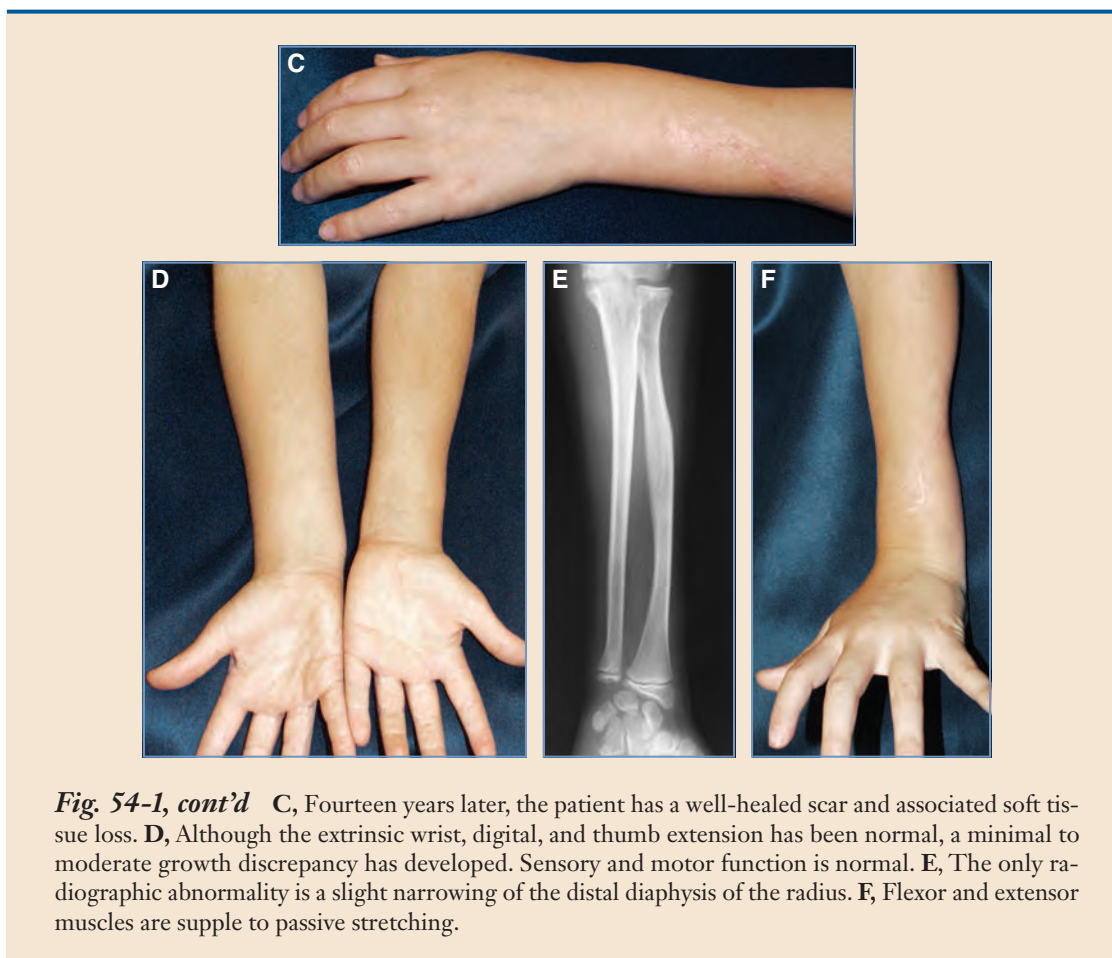
Physical Examination

Infants with Volkmann ischemic contracture are usually delivered by cesarean section. They have good Apgar scores, are neurologically intact, and respond appropriately to stimulation. Additional neurologic deficits may be seen in those with the worst deformities and include listlessness and evidence of cerebral ischemia. The most distinguishing feature of neonatal Volkmann patients is a yellow or tan eschar on the dorsal surface of the proximal forearm (Fig. 54-1). In the more severe cases, a circumferential skin eschar may be present. Extensor tightness is not typically present, because flexion contractures predominate with increasing ischemia of the flexor-pronator



Fig. 54-1 Type I defect. **A**, After a difficult labor requiring a cesarean section, this infant was born with a bullous eruption on the dorsal left forearm that persisted as a full-thickness skin ulcer by 8 weeks of age. Simple excision and closure with an advancement flap were performed. **B**, The underlying extensor tendons and muscle groups were surrounded by early scar formation within the areolar connective tissues.

Continued



muscle masses. Shortly after birth, resistance to passive extension of the flexed digits increases. Dorsal wrist and digital extension contractures develop after the skin ulcer is debrided and is either allowed to heal by spontaneous epithelialization or covered with a split-thickness skin graft. When the periosteum or denuded cortical bone is exposed, secondary osteomyelitis will occur if the open wound is not covered appropriately.

Laboratory and Radiographic Tests

Results of all routine laboratory tests are normal in infants with the milder forms of Volkmann contracture. The white blood cell count is often elevated with large ulcerating forearm lesions, and urine myoglobin levels may be elevated in the presence of extensive muscle necrosis.

Routine radiographs of the arm and forearm usually show no significant abnormalities, but as these children begin to grow, sclerotic changes with or without narrowing of the radius or ulna occur along these tubular bones at the site of bone necrosis. The mottled changes of osteomyelitis will be present with bone exposure. The distal epiphyseal ossification of both the radius and ulna is delayed in all but type I limbs. Affected growth plates become sclerotic, irregular, and mottled depending on the degree of initial vascular damage. The extent of skeletal impairment is not clear during the first several years of life, unless complete or partial absorption of the skeletal structures occurs. In patients followed through adolescence, all of these limbs with the exception of type I limbs show an impressive amount of growth retardation.

MRI studies were not carried out in any of our patients shortly after birth. Presently, special coils and techniques would be necessary to provide cuts small enough to show adequate detail in these very small limbs (see Fig. 54-4). In the future, techniques should be available to demonstrate the amount of damage to the muscle compartments.

TREATMENT (Box 54-2)

Principles

1. Early diagnosis is essential and universally overlooked.
2. Early debridement and fasciotomy are performed for all involved muscle compartments.
3. Early coverage with split-thickness grafts or preferably full-thickness flaps for large wounds is undertaken.
4. All patients undergo an aggressive splinting and passive range-of-motion regimen.
5. Stable, full-thickness coverage must be provided before elective free muscle transfers, tendon transfers, tendon lengthening, and neurolysis procedures. Waiting until all inflammation has resolved and maximum passive motion has been achieved is ideal.
6. A careful follow-up is critical to document and treat growth-related problems.

Box 54-2 Treatment Options for Neonatal Volkmann Ischemic Contracture

Muscle

Excision of muscle fibrosis before transfers

Free muscle (gracilis, partial latissimus) transplants for wrist extension or digital and thumb flexion

Classic tendon transfers for wrist extension or digital/thumb flexion, with donors depending on potential available motors (flexor carpi radialis to extensor carpi radialis to extensor digitorum communis [index through fifth], flexor digitorum superficialis [ring to thumb])

Tendon transfers for correction of fixed supination (rerouting the biceps tendon insertion)

Tendon lengthening at the forearm level

Flexor-pronator muscle slides at the forearm level

Nerve

Neurolysis at the forearm level

Nonvascularized nerve grafts (sural donor)

Vascularized nerve grafts (sural donor)

Arterial

Revascularization of the ulnar artery with a vein graft

Skin

Excision and closure of small ulcers

Local flap closure

Split-thickness skin grafts; full-thickness skin grafts

Pedicled groin or abdominal flaps

Free fasciocutaneous (anterolateral thigh, scapular) or myocutaneous (latissimus dorsi, gracilis) transfer

Skeletal

No treatment

Nonvascularized autologous or cadaveric bone grafts

Epiphyseal transfers (fibular head)

Vascularized bone grafts for nonunions (fibula, medial femoral condyle)

No distraction lengthening because of the excessive amount of scar tissue

Options

Although each affected limb in these infants is different, the early treatment for all groups is similar. Patients with these wounds, which do not heal by secondary intention, are usually referred to either a hand surgeon or a plastic surgeon for skin graft coverage. Elbow, wrist, and digital contractures in severe cases are treated aggressively with passive exercises and splinting. Infections are treated with topical and intravenous antibiotics. Numerous soft tissue and skeletal procedures have been performed in these children.

Preferred Treatment

Type I

Cleaning and local wound care are the only treatments needed in this group. Splinting and passive range-of-motion exercises may be necessary for subtle contractures within the extensor compartments.

Type II

In patients with type II, early recognition and release of tight extensor compartments will preserve muscle function. Small ulcers can be conveniently excised and closed with local or regional flap transfers of full-thickness tissue. Splinting and exercising should be started early. Progressive flexion contractures are best treated with tendon lengthening at the distal forearm instead of flexor slide procedures. We prefer to perform precise tendon lengthening at the myotendinous juncture in the forearm. The scar and secondary contracture or contractures generated by flexor slide procedures can be very troublesome. Symmetrical growth deficiencies of the forearm do not require treatment, and excessive ulnar deviation can be corrected with lengthening and bone grafting. Skeletal shortening at the forearm level will occur, often without obvious radiographic changes. No treatment is necessary.

Type III

In patients with type III, early debridement of nonviable tissue and appropriate fasciotomy of both extensor and flexor muscle groups are critical. Contracted scars from healed wounds or skin grafts should be excised and replaced with healthy, full-thickness tissue before major reconstruction is performed. Dense scars and nonfunctional fibrotic muscle (usually extensor) are best excised to prevent progressive muscle contracture. Tendon lengthening, bone grafts, and neurolysis are then carefully individualized to each limb and functional deficit.

Type IV

In patients with type IV, early debridement, fasciotomy of all muscle compartments, and revascularization of major vessels should be performed soon after birth (see Fig. 54-7). Recognition, proper diagnosis, and appropriate referral to a hand surgeon or plastic surgeon are essential. Intrinsic muscle releases are performed in swollen hands caused by major arterial blockage. Full-thickness flap replacement for heavily scarred areas should precede reconstruction for restoration of extension by free muscle transfer. Supination contractures are corrected with rerouting of the biceps tendon (see Fig. 54-5), and progressive tightness of the digital flexors is corrected by step

lengthening in the distal forearm. Repositioning of the forearm and creation of active flexion are the primary functional priorities. Well-vascularized skeletal and cartilaginous structures in the distal forearm are retained, because they may include an intact growth plate of the radius or ulna.

Type V

Early treatment of patients with type V is the same as treatment for those with type IV, with amputation of nonviable digits or thumb. All possible functional muscle should be saved, and full-thickness flap coverage of the skin-deficient areas is preferred. Neurolysis of scarred nerves and sural nerve grafting for intercalated nerve loss is performed to restore sensation in the ulnar and occasionally the median nerve distributions. When the radial nerve is so badly injured that grafting is not practical, free muscle transfer for active wrist extension is preferred to tendon transfer of scarred muscle-tendon units. Repositioning the wrist and the hand in the functional “position of advantage” is best accomplished with joint release and full-thickness skin grafting. These hands will be used as helping limbs as long as sensation is intact.

SPECIFIC TREATMENT CHALLENGES

Neonatal Volkmann ischemic contracture has no standard treatment. More patients are being correctly diagnosed and treated either at the time of birth or shortly after birth. Options for corrective surgery and/or splinting depend on the extent of the soft tissue and skeletal damage. Early treatment needs to be aggressive, because the tissue damage is always much greater than originally anticipated unless the treating physician has seen this condition previously. These accelerate with growth for all types except type I.

Patients with types IV and V losses present some of the most difficult reconstructive hand problems. Because sensation is present in these hands, they are worth salvage despite their incipient growth problems. Replacement with full-thickness soft tissue is always better than with skin grafts, which have a propensity for contracture. Free muscle transfer will substitute for the complete loss of an entire compartment, but tendon transfer in these forearms can be performed as long as the donor muscle has a good excursion and is not heavily scarred. Motor nerve identification is the most critical portion of the free transfers. Extensive neurolysis is invariably needed with remarkable return of sensation, which is not seen in adults. The hand often has intact metacarpophalangeal flexion and interphalangeal joint extension through intrinsic muscles, and thenar and hypothenar musculature is usually intact. Only the extrinsic flexors or extensors are lost. Very little can be done for patients with distal digital losses or hypoplasias. We have not performed toe transfers in these patients. Most of these children request arm and/or forearm lengthening as teens or preteens. We have performed lengthening procedures in 11 of our 49 patients over the past 40 years. The distractions have been staged, consisting of initial distraction to the desired length, followed by removal of the uniplanar device, bone grafting, and internal fixation. All have been successful, but complications were higher, including nonunion and delayed union despite bone grafting, and pin tract problems. Progress is much slower, because distraction is performed across a dense cicatrix involving soft tissue and bone.

The major goals in these severely affected limbs is to maintain motion at the shoulder, elbow, and wrist; to provide length for no other reason than appearance; and to best position the sensate hand for function. The most difficult patients are those who had intrauterine strokes affecting the uninjured side. In these patients, reconstructing the injured limb is paramount.

OUTCOMES

Information about outcomes in this group of patients is scarce.¹⁻²⁷ Most reports present the initial presentation, diagnosis, and debilitating growth disturbance. None of these children, including those with a type I limb, have completely normal forearms and hands. Those with severe neonatal ischemia have very compromised limbs. Despite the limitations, these hands are much more effective than a below-elbow prosthesis. Future reports with long-term outcomes should be forthcoming.

Type I

In patients with type I, the partial-thickness dorsal skin defects often heal spontaneously and result only in tight, hypopigmented scars. Tightness within the extensor musculature is treated with physical therapy. This is the only group without secondary growth problems (see Fig. 54-1).

Type II

In type II patients, a contracted scar or indolent ulcer on the dorsal surface of the proximal forearm can usually be excised and closed with local flap advancement. Subtle tightness within the flexor and extensor compartments can be adequately treated with splinting and stretching during the entire period of growth. Although grip strength is diminished, patients have few glaring functional impairments. Total tendon excursion at the wrist is decreased, and the tenodesis effect of the flexors and extensors with wrist range of motion can be exaggerated. Sensation is normal. Intrinsic muscle function within the hand is normal. Growth changes will result in moderate shortening (less than 25%) of the forearm (Fig. 54-2).

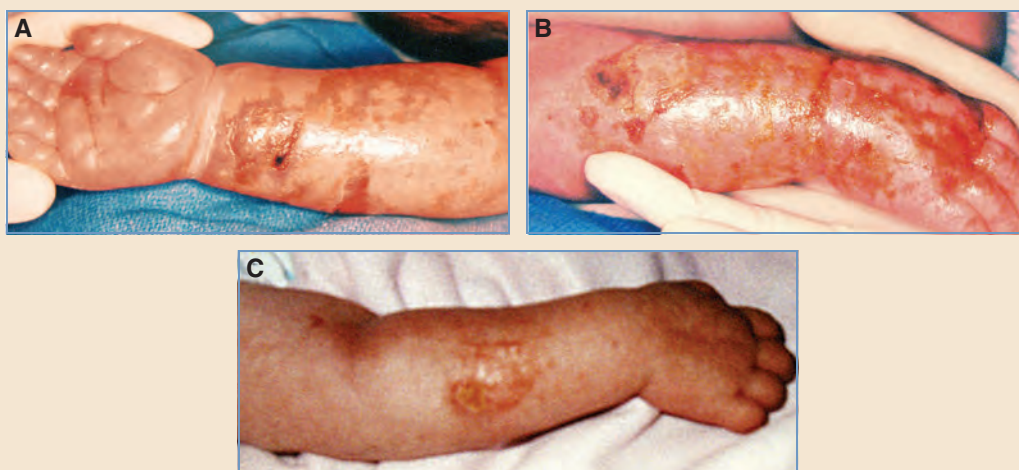


Fig. 54-2 Type II defect. **A** and **B**, After a difficult arrested labor, this infant was born with circumferential bullae involving the middle and distal thirds of the forearm. The entire forearm and hand were markedly swollen. No compartment releases were performed. **C**, A deep ulcer on the dorsal surface was the last area to heal by spontaneous epithelialization. At that time, the extensors were described as absent and the extrinsic flexor/pronator group as very tight. A splinting-stretching exercise program was initiated.

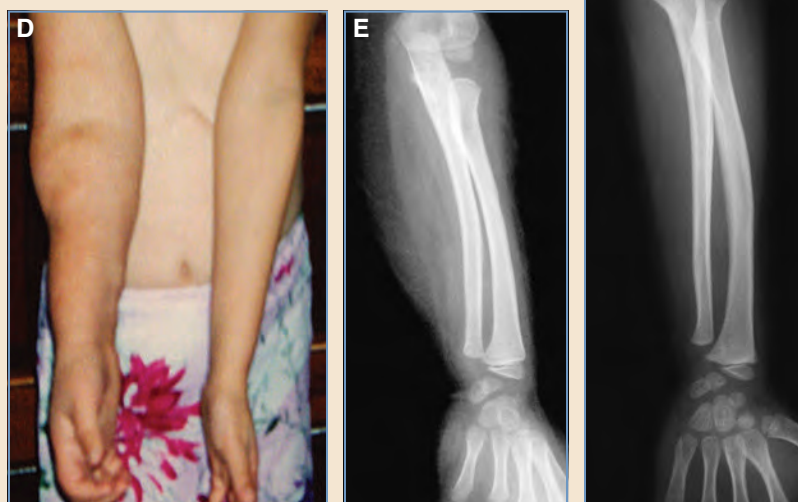


Fig. 54-2, cont'd **D**, At 9 years of age, the patient continues to have a very swollen arm with very limited wrist extension and tight extrinsic flexors of the forearm, which she cannot supinate because of the tight pronator teres muscle tendon unit. A moderate-to-severe arrest in longitudinal growth has occurred. **E** and **F**, Radiographs show no marked changes in the appearance of these tubular bones and growth plates on the right side. This forearm and hand are used primarily in a helping capacity.

Type III

Dorsal forearm wounds that either heal spontaneously or are covered with a graft will contract progressively and impressively. Aggressive stretching and splinting are more effective for the dorsal muscle compartments than for the flexor-pronator group, which often requires tendon lengthening. The hand and wrist can have marked deviation in the radial or ulnar direction with growth disturbances, resulting in forearm bones of unequal length. Osteolysis and resorption of standard corticocancellous grafts and vascularized bone grafts are increased, causing the deviation. Centralization procedures may be more predictable. The process of distraction-lengthening of short forearms when performed later in childhood and adolescence is different than distracting normal forearm bones. The large, contracted scar in the original zone of injury causes delayed skeletal unions, nonunion, and an often-difficult, slow distraction. Sensation should be intact. The vascular blood supply to the hand is intact. The moderate-to-severe shortening of the forearm bones is usually heralded by increased sclerosis and narrowing, which are seen early in childhood on routine radiographs. Tendon lengthening of the flexor-pronator group will alleviate the progressive tightness of these muscle groups and improve the gripping potential of the hand, which should have a mobile wrist. A diminished tendon excursion of the extrinsic flexors and extensors decreases the grip and causes difficulties with independent extensor function of the digits and occasionally the thumb (Fig. 54-3).

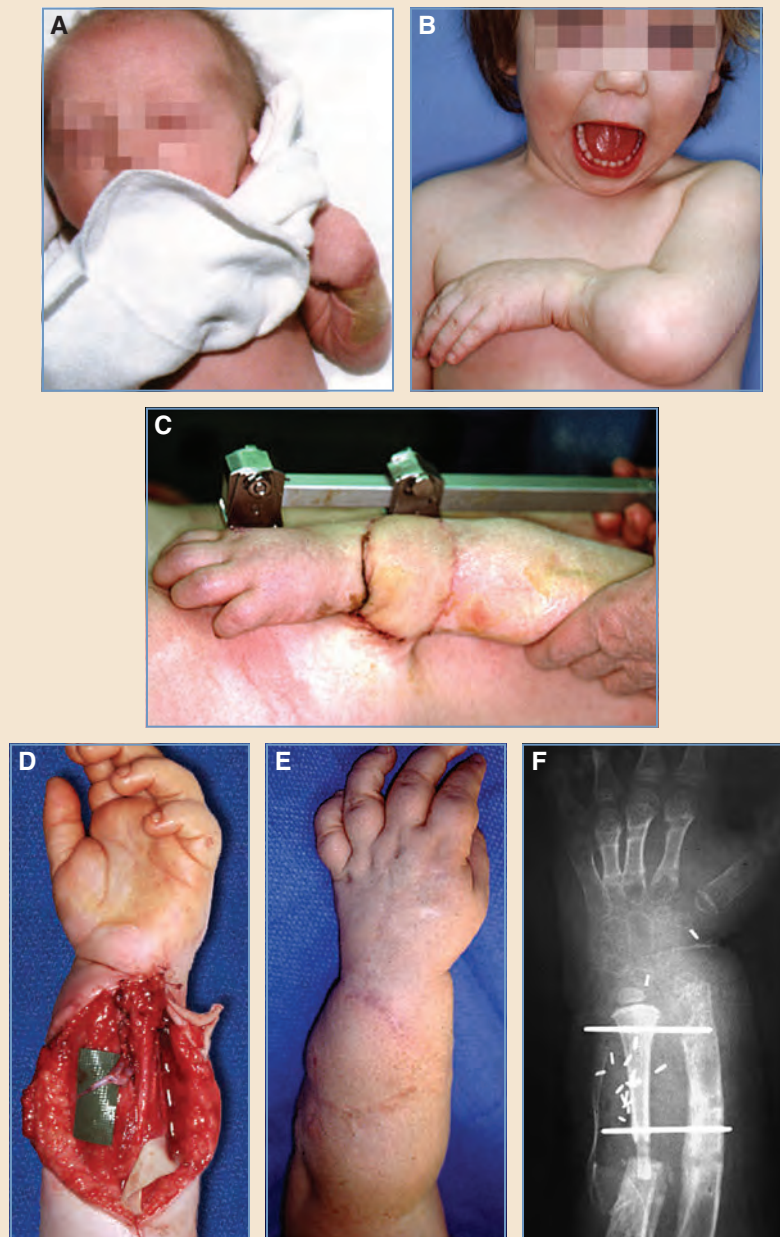


Fig. 54-3 Type III defect. **A**, This infant was delivered by cesarean section and had a yellow, parchmentlike eschar on the dorsal middle and distal thirds of the left forearm. The scab was sequentially debrided by a pediatric surgeon, who let the entire wound heal by secondary intention. **B**, She was first seen shortly after her second birthday with limited flexion and extension of the thumb and digits and severe ulnar deviation of the carpus and hand. **C**, The very tight scar on the dorsoulnar forearm was excised, and the region was resurfaced with an ipsilateral pedicled groin flap. Although sensation in the ulnar nerve distribution was intact, the ulnar artery was not in continuity. The flap was detached and inset 3 weeks later. **D** and **E**, The hand was realigned, and the loss of the distal ulna was corrected with a free vascularized fibular transfer. The peroneal artery was anastomosed as an intercalated segment between the ulnar artery in the proximal forearm and Guyon's canal distally. Additional vessels supplying the fibular head were anastomosed separately. **F**, Visible changes in the distal radius and its growth plate were evident on plain radiographs when the patient was 3 years of age.

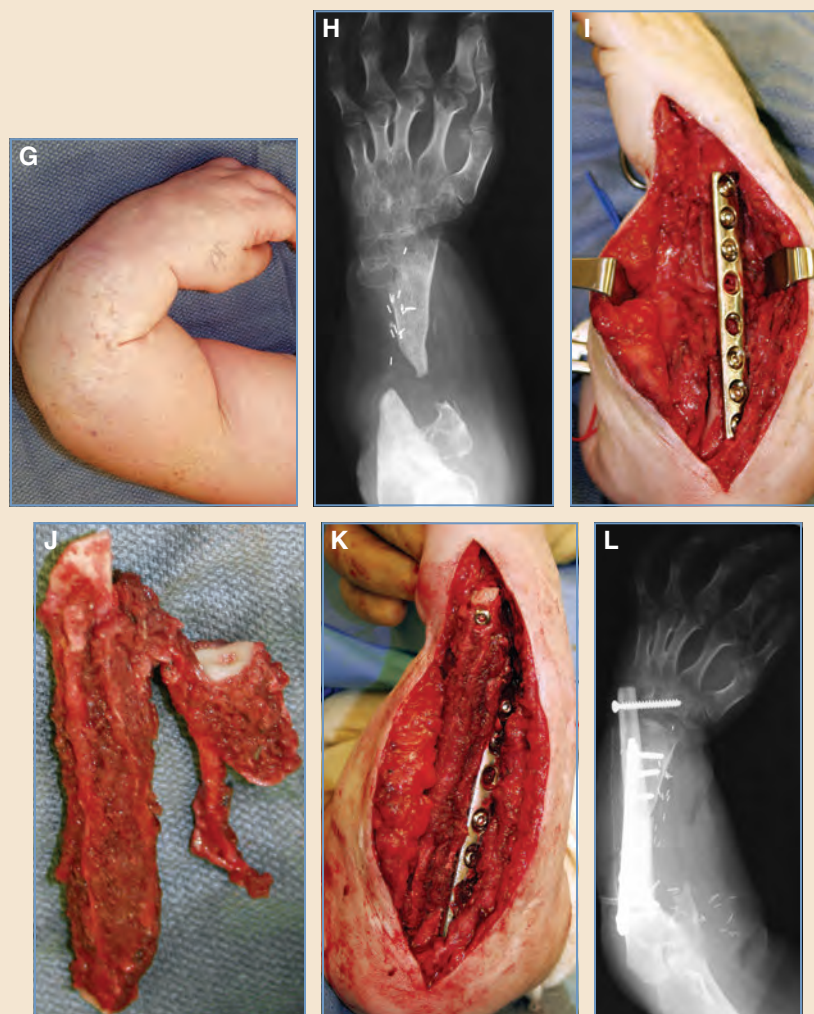
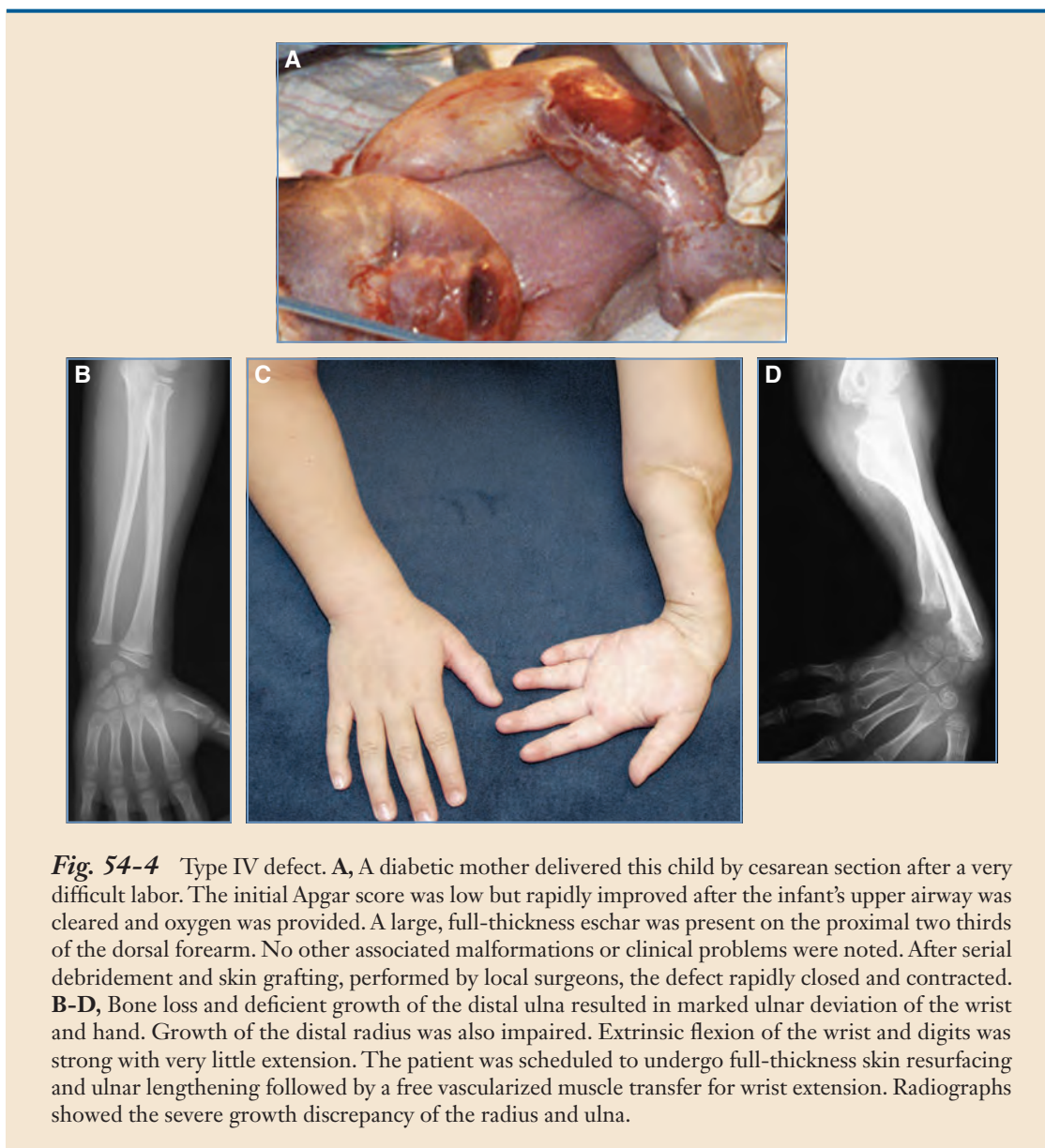


Fig. 54-3, cont'd G and H, Her floppy forearm and persistent nonunion persisted. She wore a forearm splint at all times. Sensation in the hand was normal. Ulnar-innervated intrinsic muscle function was absent. I, The longitudinal plate was reapplied, and another nonvascularized bone graft was performed. The second graft also failed to heal completely. J-L, Her last procedure was a two-segment free fibular transfer, in which one segment was placed within the nonunion, and a larger segment was placed along the entire ulnar border of the forearm. This graft has healed uneventfully and stabilized the patient's forearm and wrist. She continues to use this forearm and hand in a helping function and has always been right-hand dominant.

Type IV

All of the adverse outcomes seen in the type III category are predictable in patients with type IV. The arms, wrists, and hands of these patients are never normal. Skeletal changes are apparent early in childhood, and the diminution of growth is severe. Distraction lengthening is difficult. Distal ischemic changes in the hand can result in partial or complete digital loss. One patient with radial artery loss and an incomplete palmar arch had a complete thumb loss (see Fig. 54-5). Digital and thumb range of motion is often affected, and grip, key pinch, and independent precision function of the hand are affected to varying degrees. These children adapt to their functional



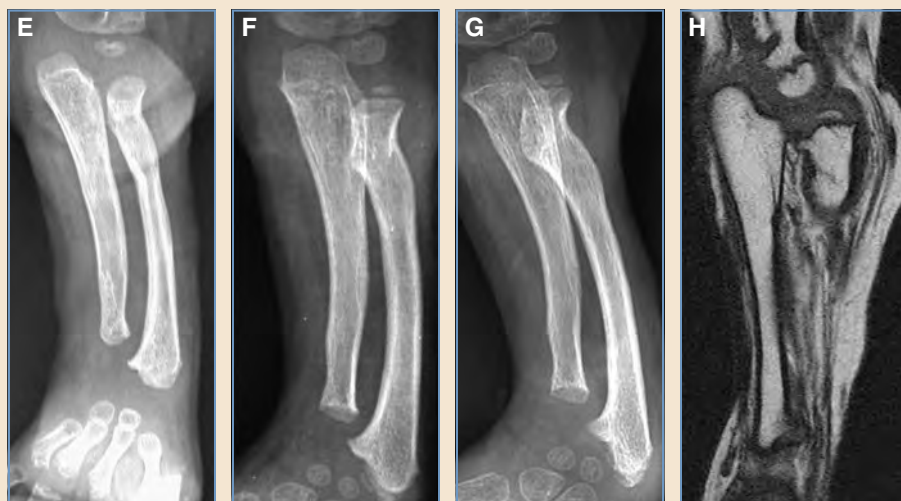


Fig. 54-4, cont'd E-G, Serial forearm radiographs obtained at 4 months, 2 years, and 7 years of age demonstrated early epiphyseal mottling, irregularity, and very deficient growth of the distal ulna and radius. H, Sagittal MRI at 7 years showed marked loss of skin, subcutaneous fat, and all local muscle groups.

deficits and develop many ingenious compensatory motions just as those with congenital malformations do. The results of early neurolysis of scarred nerves or intercalated nerve grafting are excellent for return of protective sensation. The return of intrinsic muscle function in the hand in these situations is unpredictable. Those with one-vessel forearms may demonstrate varying degrees of cold intolerance in colder northern climates. Many of these children do not complain, even though the affected forearms and hands become quite mottled and erythematous when exposed to the cold. This is not unusual in these children, who have adapted to both their lack of mobility and altered sensation since birth. This is normal to them. Because the cicatrix from the original zone of injury is much greater, outcomes after muscle slide and tendon lengthening are poor to fair at best. Free muscle transplantation is more predictable (Fig. 54-4).

Type V

Type V limbs function as helping extremities at best. Young children often neglect digits without sensation, which are subject to injury, maceration, and infection. The initial wounds are often circumferential and result in tight, contracting scars early in life. Often, major portions of potentially functional muscles and their motor nerves are debrided. Bone loss, usually ulnar, frequently results in severe deviation at the wrist level. The motion of most joints within the hand and wrist is affected, and growth is frequently diminished. The digits and thumbs with intact sensation and good extrinsic and intrinsic muscle function will grow most predictably.

Despite the devastating effect of these ischemic injuries involving almost all skeletal and soft tissue structures of the forearm, these children adapt very quickly to their deficiencies and make the best of potential function (Figs. 54-5 through 54-9).

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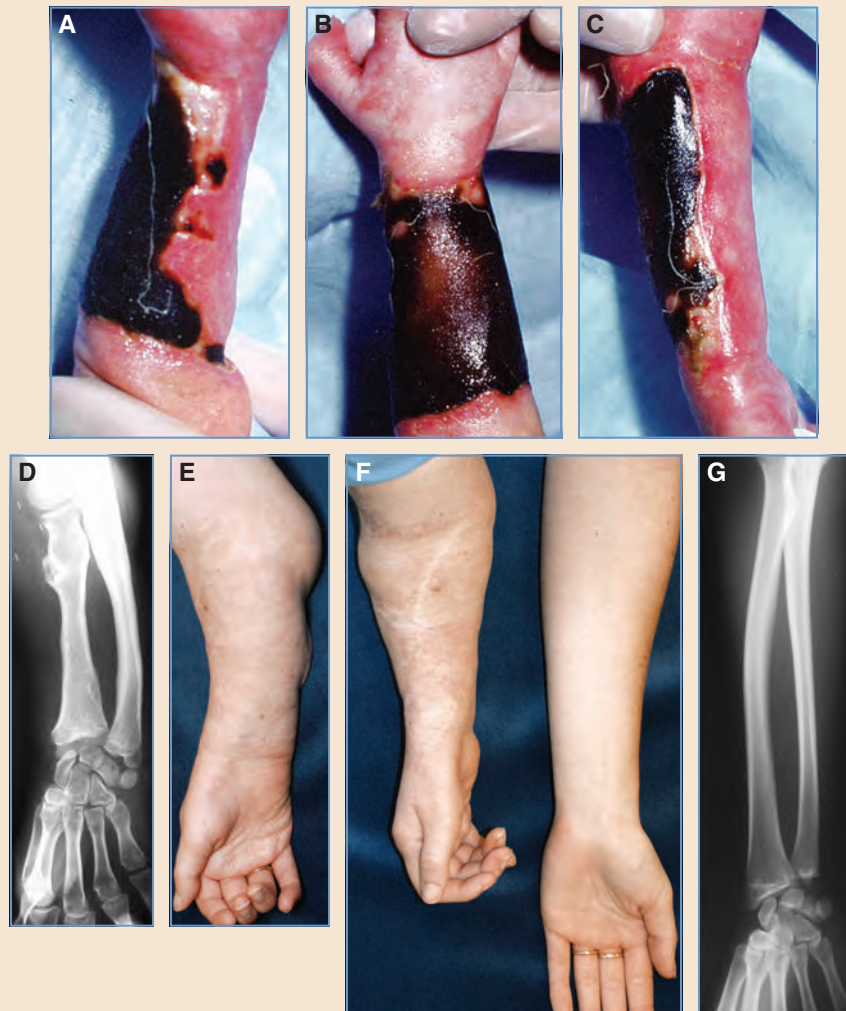


Fig. 54-5 Type IV defect. **A-C**, This infant was born vaginally after a very difficult and prolonged labor. By 2 weeks of age, she had a full-thickness eschar. The mother had gestational diabetes and toxemia of pregnancy. The wound was debrided over the next 3 weeks, and the granulation bed was covered with a split-thickness skin graft by a pediatric surgeon. **D-G**, The clinical appearance and radiographs of the patient's right and left forearms and hands at 22 years of age. The reconstruction consisted of pedicled flap coverage of the entire dorsal forearm at 3 years of age, a free gracilis muscle transfer for wrist extension at 7 years of age, an osteotomy and lengthening of the radius at 9 years of age, and re-routing of the biceps tendon for correction of a fixed supination contracture. She has had normal sensation, intact radial and ulnar arteries, and tight flexor tendons for her entire life.



Fig. 54-6 Type V defect with distal digital loss. **A** and **B**, This 6-year-old child was born with a circumferential eschar that was debrided and skin grafted within the first 3 months of life. The avascular thumb was amputated. Despite several releases, the severe extension contractures have persisted; in addition, the patient had secondary boutonnière deformities involving all four remaining digits. This hand is minimally functional. **C** and **D**, MRA scans showed normal trifurcation of the brachial artery at the elbow. Only the ulnar artery was in continuity. The interosseous and radial arteries were blocked at the distal forearm level and reconstituted with collateral circulation to fill the common digital vessels to the digits. There was no evidence of either an intact superficial or deep palmar arch.

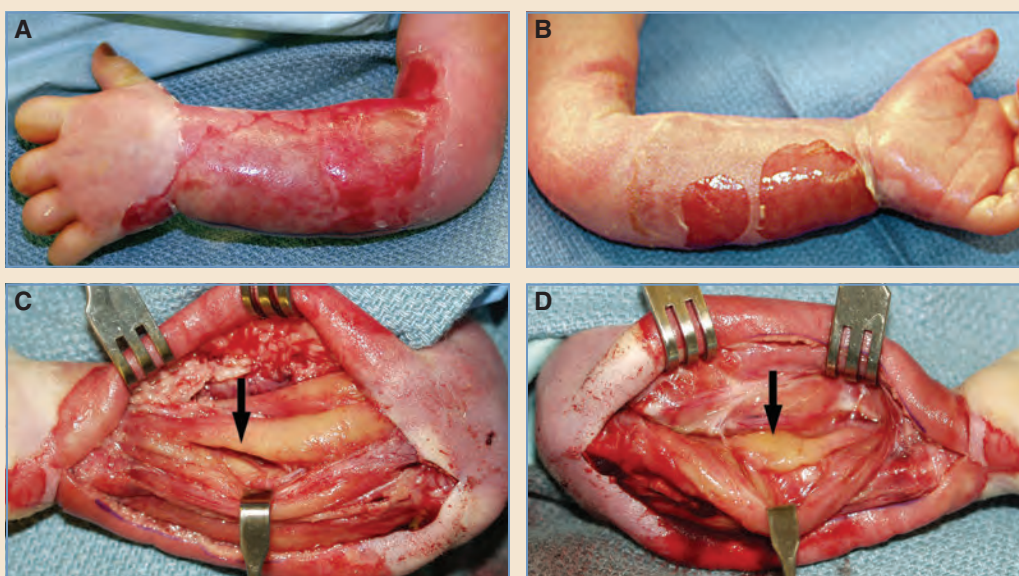


Fig. 54-7 Type V defect with dorsal and volar compartment loss. **A** and **B**, This child's mother reported abnormal fetal movement 2 days before giving birth by cesarean section. The child, a dizygotic twin, was born with circumferential blistering around the entire forearm and is shown at 2 days of age. There was a 22-hour delay in making the proper diagnosis. By that time, the entire hand and all of the digits were swollen and very painful to passive extension. **C** and **D**, In a forearm and hand fasciotomy, nonviable lemon-colored muscle was observed in the dorsal and volar compartments (*arrows*). The maximal compression was located on the dorsal surface of the proximal and middle thirds of the forearm. None of the blistered skin survived.

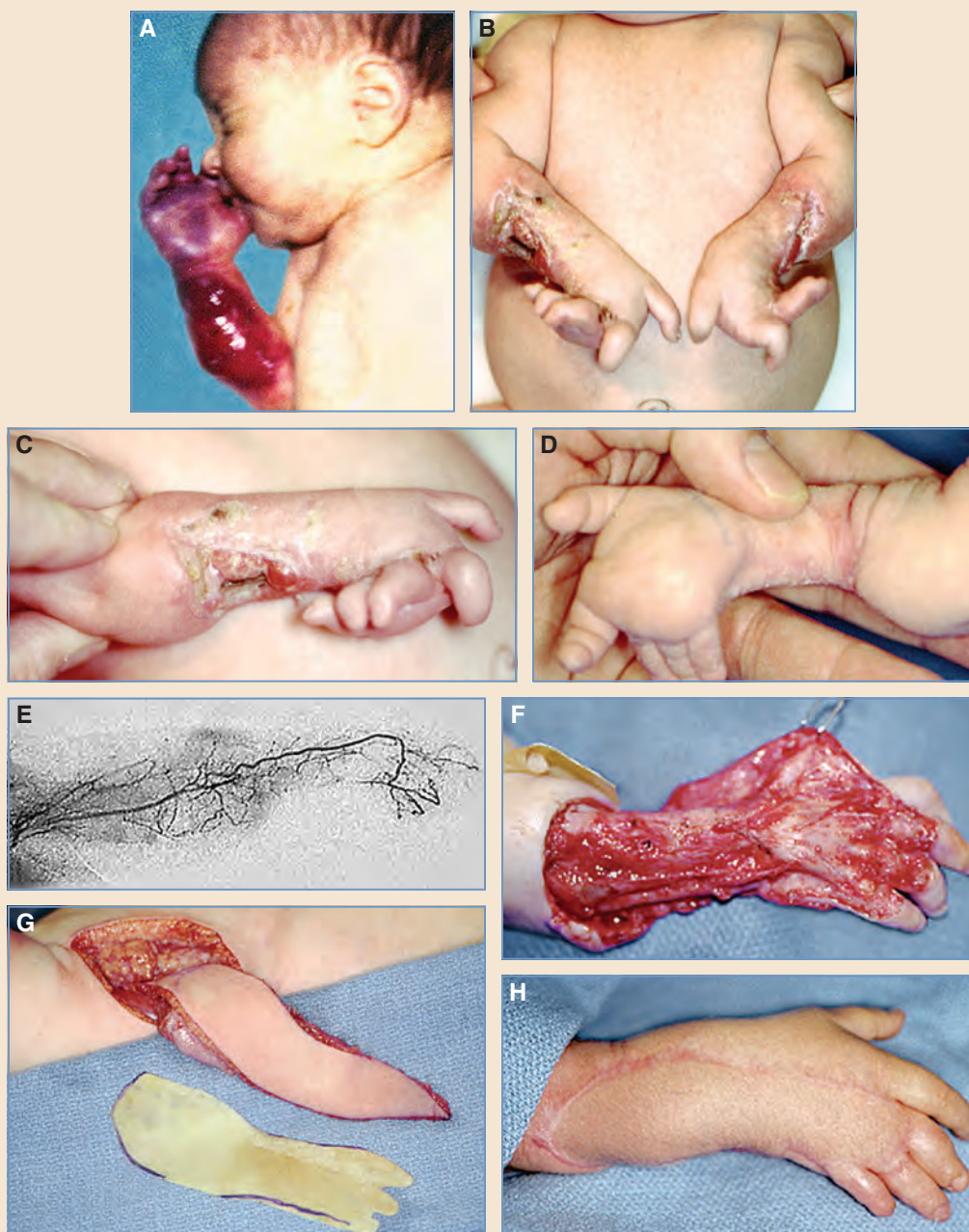


Fig. 54-8 Type V defect with allograft transplantation. **A**, This monozygotic twin was born by cesarean section, with circumferential discoloration and bullous eruptions on both forearms. The child had severe vascular compromise of the ulnar half of both hands for the first 3 weeks of the life. **B**, When first seen at 5 weeks of age, the patient had large, open ulcers of the dorsal and ulnar surfaces of both forearms, along with exposed tendon and bone. **C** and **D**, Impressive contractures and extensive soft tissue loss had already developed. A 6 cm area of ulnar diaphysis was exposed and infected. **E**, A preoperative angiogram revealed a brachial artery feeding a one-vessel (ulnar artery) forearm and an intact palmar arch within the hand segment. **F**, The extent of the soft tissue skin and muscle loss was appreciated after scar debridement, fasciotomy of all dorsal and volar compartments, and contracture release. No normal skin was debrided. **G** and **H**, The forearm, wrist, and hand were covered with a free latissimus dorsi myocutaneous flap at 6 weeks of age. The thoracodorsal artery was anastomosed end to side to the 1.5 mm brachial artery at the elbow level.



Fig. 54-8, cont'd **I-K**, A large intercalated defect within the ulna remained after multiple debridements. The distal ulna was originally intact, with vascularized cartilage with a growth plate. Shortly after the infant's father tragically died of a ruptured intracerebral aneurysm, a portion of his radius and radial nerve was transplanted as allografts to reconstruct the ulna and ulnar nerve. She was maintained on a low-dose immunosuppressant for 10 months after the transplantation. **L-N**, At 20 years of age, the free latissimus flaps used to resurface both forearms have expanded with growth, which is impaired on both forearms. Although longitudinal growth of both radii exceeds that of either ulna, forearm length is markedly diminished. **O**, The twins at 19 years of age, with the affected woman on the *right*. Thumb and index finger pinch are good in both hands. She has no wrist extension or digital extension of the ulnar three digits, which show evidence of vascular impairment. Thumb and index finger extension are present. Light touch and 4 mm, moving, two-point discrimination are present in both ulnar nerve distributions.

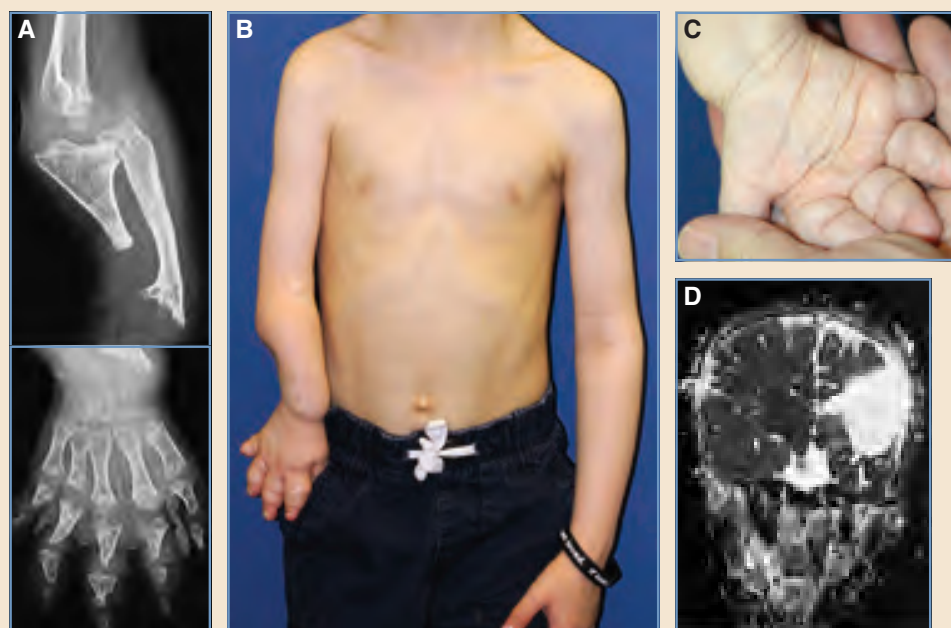


Fig. 54-9 Type V defect with stroke. **A** and **B**, This child presented at 5 years 4 months of age with a marked length discrepancy of his right upper limb. He was born with a major ischemic injury and a large eschar on the dorsoulnar surface of the forearm, which healed spontaneously. The shoulder was high positioned. The elbow and wrist were unstable. He had good shoulder and elbow motion. The wrist was flail. Radiographs showed that, with growth, the distal ulna completely resorbed, and the radius became sclerotic with a deformed, angulated distal metaphysis. The phalanges had thin, pencil-like condyles and hypoplastic distal segments. **C**, The hand appeared swollen and was much smaller than the opposite hand. Although the hand wrinkled in water, he had diminished sensation in the medial nerve distribution. **D**, An MRI T2-weighted scan showed an ischemic defect in the distribution of the middle cerebral artery.

Childhood Volkmann Ischemic Contracture

The childhood form of Volkmann ischemic contracture is a clinical condition resulting from muscle necrosis and subsequent secondary shortening and contracture. The muscle necrosis and loss often lead to a significant functional deficit. This, combined with tightness and contracture formation, can reduce hand function to a minimum. Volkmann²⁴ first described the condition in 1869. Classically, the contracture follows a supracondylar fracture. Circulatory embarrassment occurs, which can lead to compartment syndrome, as described later in the chapter. The end result is a pronated forearm with the wrist flexed, the thumb adducted, and the metacarpal phalangeal joints extended. The interphalangeal joints of the fingers and thumb are flexed. Most cases of childhood Volkmann ischemic contracture can be prevented by appropriate management of the supracondylar fracture and early recognition and treatment of developing compartment syndrome. Once established, however, the management becomes extremely complex and difficult. In severe cases, a variety of reconstructive techniques, including microvascular functioning muscle transplantation, are possible. However, even with the sophisticated reconstructions available today, normality can never be restored. Thus, prevention is paramount, and additional, more effective management techniques should be sought.

CAUSES AND PATHOGENESIS

Knowledge of the anatomy of the forearm is helpful for understanding the causes and pathogenesis of Volkmann ischemic contracture. Most cases result from a supracondylar fracture with a subsequent untreated or inadequately treated compartment syndrome. The forearm musculature is encased in a thick fascial envelope. With decreased perfusion, the enclosed musculature swells. However, the fascial envelope limits swelling and leads to further ischemia. A vicious cycle of increasing ischemia begins, leading to further edema and reduced circulation. Eventually, circulation in the muscle is severely diminished, resulting in muscle necrosis. This reduced arterial input is known as *Volkmann ischemia* and is basically a process of myonecrosis. If unrelieved, irreversible muscle death will occur. To relieve the pressure, the compartment is opened through fasciotomies. The ischemic muscles rapidly tighten and shorten. Over the course of several weeks, the forearm and hand take on the classic characteristics of Volkmann ischemic contracture. In severe cases, the tight volar forearm musculature tightens the tendons, deforming the fingers and resulting in an immobile hand of minimal functional value.

Lesser degrees of involvement occur, but most cases are severe. For children and older (not neonatal), Lipscomb²⁶ has classified Volkmann ischemic contracture into four categories according to the severity. Grade I is mild with good muscle function that is only minimally reduced. Nerves are not involved. Grade II is moderate with considerable loss of muscle function but no nerve deficit. Grade III is severe with only minimal residual muscle function. Some degree of nerve deficit is typical. Grade IV is severe with no muscle function and significant nerve deficit. From a clinical standpoint, most treated cases are grade III or IV. Some patients with grade I are not recognized.

PREVENTION

Volkmann ischemic contracture is a condition that should not occur in modern surgical practice. Appropriate treatment of supracondylar fractures and early recognition and treatment of any suspected compartment syndrome are essential. Surgeons should be on alert with high-risk patients, such as those with displaced supracondylar fractures. Managing these fractures involves closed reduction with fluoroscopic control and insertion of percutaneous K-wires. The reduction is maintained with the K-wires, and the elbow can then be extended, thus minimizing the chance of reduced arterial flow in the brachial artery. Even with appropriate reduction, adequate fixation, and the elbow in extension, extreme vigilance is needed to recognize the signs and symptoms of a compartment syndrome. Monitoring children for symptoms that are cause for concern is imperative. These include increasing pain in the forearm, reduced sensation in the hand, reduced arterial input to the hand, and increased swelling and tenseness in the forearm compartments. Surgeons should monitor the vascularity through the radial and ulnar pulses and watch for increasing forearm discomfort, particularly with passive extension of the fingers. With passive stretching of the fingers, the musculotendinous units are placed under some degree of tension and will be extremely painful if an ischemic process is under way. Flexing the fingers will be extremely painful if forearm ischemia is present. If compartment syndrome is suspected, the tense, intracompartmental pressure is measured. This is easy to perform at the bedside with a wick catheter or a slit catheter technique. If the intracompartmental pressures exceed 30 mm Hg, decompression by fasciotomy is indicated.

Fasciotomies are carried out as soon as possible and as completely as possible. Volkmann ischemic contracture can involve not only the volar compartments, but also the dorsal compartments. Thus these are monitored and decompressed if necessary. Early decompression can interfere with the vicious cycle of muscle ischemia, swelling, and necrosis. However, when muscle is ischemic

at room temperature for longer than 6 hours, a full recovery is not possible.^{27,28} Thus timing is critical, and compartment syndrome release should be considered a high-priority emergency. The fasciotomy wound is left open and closed later, when muscle swelling has diminished. The wound can be closed with a variety of skin closure devices or a skin graft. Occasionally, the skin graft can be excised at a later date with the aid of tissue expansion. Although the forearm can scar after this is done, upper limb and hand function are preserved.

MANAGEMENT OF ESTABLISHED VOLKMANN ISCHEMIC CONTRACTURE

The goal of managing an established Volkmann ischemic contracture patient is to restore as much function to the extremity as possible. A thorough patient evaluation is performed, and reconstruction of the injured tissues is planned.

Clinical Assessment

A detailed history is obtained, including previous surgical procedures, with particular attention to surgical debridement. Distinguishing the involved nerves is difficult when the tissues are necrotic. These have at times been mistakenly debrided. The functional impairment as perceived by the patient and family is documented. A detailed physical examination is performed that includes all structures in the forearm and hand from the circulatory system to the nerves, muscles, joints, and soft tissues. As noted in Table 54-3, the arterial input should be assessed by brachial, radial, and ulnar pulses, as well as angiography. An angiogram can provide valuable information about the anterior interosseous vessels. These are the key vessels involved in muscle transplantation. They lie beside the key nerve that will innervate the muscle, namely, the anterior interosseous nerve. Thus an angiogram is useful not only for assessing the continuity of the brachial, ulnar, and radial arterial systems (Fig. 54-10), but also for obtaining valuable information about previous damage to the anterior interosseous nerve. If an angiogram shows a healthy, pristine anterior interosseous artery, then the anterior interosseous nerve is probably uninjured. The vessels will be used as the recipient vessels in a muscle transplant procedure, and the nerve will be used as the motor nerve to innervate the muscle transplant.

Table 54-3 Clinical and Laboratory Assessment

Structure	Clinical Examination	Laboratory Evaluation
Arterial input	Pulses and Doppler	MRA and angiogram
Venous outflow	Observation and Doppler ultrasound	Angiogram
Nerves	Median, ulnar, and radial nerve testing	EMG Nerve conduction studies EMG of pronator quadrates
Muscle function	Testing of each muscle	EMG
Joint function	Active and passive range of motion	Radiographs
Soft tissues	Scar limits movement Provision for gliding	MRI



Fig. 54-10 An angiogram showing the anterior interosseous vessels.

The major nerves to the forearm are assessed. A clinical examination of the median, ulnar, and radial nerves may be aided by EMG and nerve conduction studies. Very often, diminished nerve function is related to an ischemic process that has involved the nerves, similar to the process of myoneural necrosis. When the nerve is heavily encased in scar, it is sometimes helpful to release the scar with a neurolysis to provide additional motor and sensory function in the distal component of the extremity. When median and/or ulnar nerve function is absent, a physical defect in the nerve should be suspected and reconstruction considered. Motor nerve function can also be evaluated by EMG of the pronator quadratus muscle. This muscle lies distal in the volar forearm; therefore, if its function is confirmed, it is very likely that the anterior interosseous nerve will be functional. They are both branches of the median nerve, and some degree of active EMG activity is indicative of a functioning nerve even in the face of extensive muscle damage from the ischemic process. If uncertainty persists regarding the function of a needed motor nerve, a nerve biopsy may be helpful. This becomes particularly important in patients with a high arm or axilla injury.

Nerve reconstruction is probably best performed before motor reconstruction, with the aim of obtaining a sensate hand. Often, nerve grafting is needed to correct a significant nerve gap. All muscle groups are carefully evaluated. Even a flicker of motion is important to document. This often indicates that even though most of the muscle is necrotic, the motor nerve to the muscle is functioning. This can be crucial information when planning a muscle reconstruction.

Adequate mobility of the wrist and fingers is essential. This may be limited by the tightness of the contracture, and attempts should be made to improve the range of motion through rehabilitation. If the tendons are tight, it may only be possible to achieve partial joint mobility, but surgeons should attempt to improve this as much as possible. It is futile to provide a new motor nerve to a joint if the joint is stiff. Thus it may be necessary to surgically release tight, immobile joints before a muscle reconstruction is attempted.

The presence of extensive tightness in the volar soft tissues should be fully evaluated (see Table 54-3). Soft tissue tightness can not only limit passive joint mobility, but also impinge on muscle contraction and tendon gliding. Thus it may be necessary to provide an adequate soft



Fig. 54-11 A pedicled flap in preparation for a muscle transplantation.

tissue cover and release all soft tissue tightness. This can be accomplished with a distant pedicle flap (Fig. 54-11) or a free tissue transfer.

Patients with Volkmann ischemic contracture should undergo a psychosocial assessment. The child and family should be given realistic expectations regarding reconstruction and a detailed outline of the postoperative requirements. A well-motivated patient is essential. A rather complex and time-consuming rehabilitation program will be needed to ensure maximal gain from the reconstruction. Considerable rehabilitation will be necessary; therefore the patient needs to be compliant and the family prepared to work with the child during the postoperative rehabilitative phase. Otherwise, the gain will be only minimal.

Preoperative Procedures

A variety of factors are considered before a reconstruction of a Volkmann ischemic contracture. First, the vascular input needs to be addressed. A defect in the brachial artery should be corrected to achieve maximal inflow to the forearm and hand (Fig. 54-12). A sensate hand is important to achieve before any form of motor reconstruction is attempted. Thus median and ulnar nerves need to be in continuity, with their functional potential optimized. This may involve neurolysis, nerve grafting, or nerve transfer. The joints that will be activated by a muscle reconstruction need to have adequate range of motion. Capsulotomies may be necessary before motor reconstruction is undertaken. The soft tissues should be in optimal condition to provide adequate muscle contraction and tendon gliding.

Preoperative physiotherapy is often helpful to achieve as much mobility to the forearm and hand as possible. Pronation and supination of the forearm are emphasized. This is frequently a very difficult problem to address after muscle transplantation. If mobility is not possible, then the forearm should be placed in the most functional semipronated position. Attempts should be made to improve forearm pronation and supination. To do this, a tight pronator teres muscle may require release. In addition, the pronator quadratus in the distal forearm is often tight and limits supination. The next structure that can be involved is the interosseous membrane. This may require an incision to provide passive supination. Last, the tight capsule around the head of the radius might need to be released. Even with these four procedures, the amount of active supina-

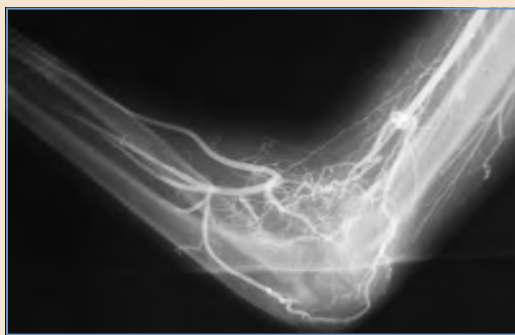


Fig. 54-12 An angiogram reveals a defect in the brachial artery.

tion and pronation gained is often disappointing. However, even a small amount of movement is worthwhile and will provide a base on which to build.

When the circulation optimized, the sensory input is optimized, and the joint and soft tissues are fully mobilized, then additional active muscle function can be provided. Free-functioning muscle transplantation to the forearm has revolutionized the care of childhood Volkmann ischemic contracture.^{29,30} Although far from providing normal function, it can upgrade the usefulness of an extremity enormously. This procedure is the backbone of reconstruction of severe Volkmann ischemic contracture in childhood and is described in detail in the next section.

Muscle Transplantation

History

The onset of microsurgical free tissue transfer ushered in a new era of reconstructive surgery. It was not long before researchers and microsurgeons recognized the possibility of functioning muscle transplantation. In this procedure a muscle is transplanted to a different part of the body and revascularized as in other free tissue transfers, and it is reinnervated and appropriately positioned so that it will function. In 1970 Tamai et al³¹ reported on a successful transplantation of the rectus femoris muscle to the forelimb in a dog model using microneurovascular techniques. Both electrophysiologic and clinical evidence of muscle contraction and function were presented. This formed the basis for ongoing research and development in this field. In 1973, at the Sixth People's Hospital in Shanghai, microsurgeons transplanted a lateral portion of the pectoralis major muscle in a patient with Volkmann ischemic contracture.³² This was the first successful clinical application of functioning muscle transplantation. Remarkably, the patient achieved a good range of finger motion and substantial grip force. Kubo et al³³ carried out further confirmatory research in 1976, showing a virtually normal histologic appearance after muscle transplantation. In 1976 Harii et al³⁴ documented the role of functioning muscle transplantation for the rehabilitation of facial paralysis. They transplanted the gracilis muscle to the face with innervation from the healthy ipsilateral motor component of the fifth nerve.

Ikuta et al³⁵ reported on the reconstruction of Volkmann ischemic contracture by functioning muscle transplantation in a 6-year-old child. Manktelow et al³⁶ performed a functional transfer to the forearm using the gracilis and pectoralis major muscles. In 1991 Zuker et al³⁰ described their results of functioning muscle transplantation in Volkmann ischemic contracture in children. In seven patients the gracilis muscle was used for forearm reconstruction. All seven children achieved

independence in terms of daily living and significant functional improvement, particularly relative to static grip and pinch patterns. Some, however, had persistent difficulty with fine motor function, which was largely related to a lack of intrinsic hand capabilities. All children had limb length discrepancies despite open growth plates. Numerous other investigators have demonstrated an improvement in function to the forearm and upper extremity after functional muscle transplantation.³⁷⁻⁴³

Indications

Muscle transplantation for Volkmann ischemic contracture is indicated when a significant functional gain can be achieved with the addition of a single functioning muscle.^{44,45} Often, the extensor compartment is damaged, and the muscle is weakened and unsuitable for a tendon transfer. Transplantation of a muscle from a distance can significantly improve function, particularly that related to long finger grip. As with tendon transfers, the prerequisites for muscle transplantation involve adequate proximal stability of the limb, adequate ability to appropriately position the hand, and a virtual normal range of passive mobility of the wrist, fingers, and thumb. At a minimum, protective hand sensibility and preferably normal sensibility should be present. Last, the soft tissue should be adequate to provide tendon gliding at the site of tendon repair in the distal forearm. Equally important to all of these physical prerequisites is a well-motivated, determined patient with sufficient resources for intensive follow-up and postsurgical rehabilitation. If the physical and psychosocial prerequisites are met and a single, functioning muscle will significantly improve function, a muscle transplant is indicated.

As mentioned previously, other procedures may be necessary before a functioning muscle transplantation is performed. These will optimize the early and late success of the transplantation. Circulatory disruption should be repaired. A damaged brachial artery may require grafting to improve the arterial input to the forearm. Nerve gaps need to be corrected by either direct coaptation or nerve grafting. A minimum of protective sensibility should be present in the hand to improve function with a muscle transplant. Muscle contraction cannot move immobile joints. Thus these need to be addressed and adequate passive range of motion achieved. Tendon gliding is optimized with healthy flap coverage in the distal forearm (see Fig. 54-11). In addition to all of these physical prerequisites, enrolling the child in a detailed rehabilitation program may be helpful. Concepts of what will be needed after surgery should be provided and realistic goals set.

Surgical Technique of Muscle Transplantation to the Forearm

The surgical procedure for reconstruction of the forearm musculature after Volkmann ischemic contracture is difficult and complex. A two-team approach is best. The gracilis muscle is ideal for such a transfer from both an anatomic and physiologic perspective.⁴⁶ The concept of functioning free muscle transplantation in various locations in the upper extremity is similar in principle and is described here for the finger and thumb flexors. These principles can be easily applied to the finger and thumb extensors, the elbow flexor (biceps), and the shoulder abductor (deltoid).

The surgical preparation of the forearm can be difficult and tedious because of extensive scarring and previous surgical debridement. The incision along the volar forearm should be carefully planned. In the proximal forearm, the incision should adequately expose the medial epicondyle and the neurovascular recipient structures. In the distal forearm, skin flap coverage will be required for coverage of the tendon repairs to facilitate tendon gliding (Fig. 54-13, A).

Generally, beginning the dissection proximally where healthy undamaged tissue can be identified is preferred. The dissection continues distally using the median nerve and brachial artery as a guide for the depth of dissection. Preserving any large superficial vein in the proximal forearm is helpful, because it may be preferable to a vena comitans of the deeper vasculature. For finger and thumb flexion, the best nerve to use is the anterior interosseous nerve, which is a branch of

the median nerve. Adjacent to its takeoff from the median nerve are the anterior interosseous vessels, which can also provide an excellent vascular supply for the transplanted muscle. The anterior interosseous vessels and nerve are exposed by transecting the insertion of the pronator teres. This is reflected to reveal the neurovascular bundle. Even in severe cases of Volkmann ischemic contracture, stimulation of small areas of residual musculature may be possible. Using the nerve stimulator on the anterior interosseous nerve, residual musculature may contract somewhat. This will not be effective clinically, because it does not produce movement. However, it will confirm the viability and functionality of the anterior interosseous nerve. Thus this maneuver can be very important and very reassuring to surgeons.

Once the anterior interosseous nerve has been clearly identified and the anterior interosseous artery prepared, attention is turned to the central and distal forearm. The skin flaps should be elevated in a fashion that ensures no tension on the muscle with closure. In the incision in the distal forearm, it is particularly important to provide a skin flap for coverage over the tendon repairs. As the distal forearm is entered, the profundus tendons are identified. The superficialis tendons can often be divided, because they are not useful in this procedure. The profundus tendons are divided and then sutured to one another in a balanced fashion. In this way, when traction is applied to the tendons, the fingers move down into the flexed position in unison. The flexor pollicis longus is identified and divided in the forearm. If it is to be incorporated into the single muscle transplant, then it should flex slightly after the fingers. This ensures the thumb will not be caught in the palm and will not decrease the effectiveness of grip. It should flex after the fingers to provide thumb-index finger apposition. The flexor pollicis longus is then sutured to the

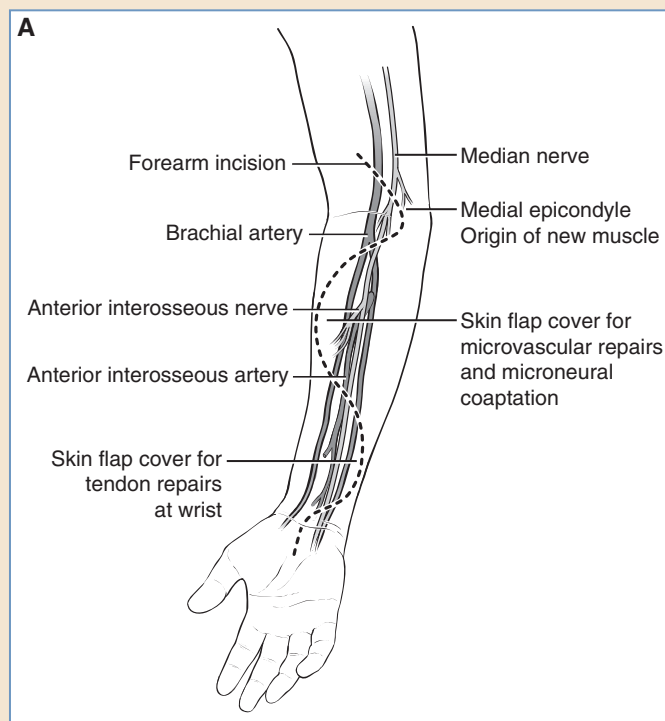


Fig. 54-13 A, A volar forearm incision.

balanced, four profundus tendons, and the preparation of the forearm is complete. Much of this dissection can be done under tourniquet control. However, an adequate arterial supply for vascularization of the muscle should be confirmed as the tourniquet is deflated. With the tourniquet deflated, a functioning anterior interosseous nerve is confirmed by stimulation and observation of contraction of the residual muscle.

In unusual cases in which the anterior interosseous nerve is not available, branches of the ulnar nerve that innervate the ulnar profundus tendons may be used as a second choice.

In some situations, having a degree of independence of long finger flexion and thumb flexion is helpful. This may be possible with the use of the anterior interosseous nerve. This nerve courses distally, giving off separate branches to the profundus and separate branches to the flexor pollicis longus. The branches that go to the profundus course ulnarly. The branches that go to the flexor pollicis longus course radially.⁴⁷ Thus the motor components for finger flexion can be separated from those for thumb flexion. It may be possible to separate the fibers according to fascicular territory when transplanting the gracilis muscle. In this way, the surgeon can create a separate longitudinal section of muscle with its tendinous unit, innervated by a separate fascicle. With appropriate positioning and identification of the motor components and tendinous components, the thumb function and the finger function can be separated to some degree (Fig. 54-13, B).

The volar forearm is now fully prepared. The medial epicondyle has been exposed, and this will anchor the muscle origin. The motor nerve to be used has been carefully prepared and confirmed to have adequate function. Often, a neuropathologist can confirm the presence of healthy axons by a quick section evaluation. Adequate vascular input is ensured by releasing the tourniquet and testing the arterial flow. A superficial vein is often helpful where a deep vein is not present or is involved with extensive venous communications. In the distal forearm, the four profundus tendons are divided and sutured in unison with a side-to-side woven technique. Traction on this unit produces finger flexion in unison. The flexor pollicis longus may be incorporated into the profundus tendon if a single, nonsplit gracilis transplant is to be carried out. The thumb is incorporated. It should lag slightly behind the unified finger flexors. If a split-gracilis muscle transplant is planned, then the anterior interosseous nerve is appropriately split and dissected, and the finger and thumb flexors are left separate.

Gracilis Muscle Preparation

Using a two-team approach, the muscle is harvested simultaneously with the forearm preparation. Many muscles have been used for transplantation, but the gracilis appears to be the best choice for forearm reconstruction because of its anatomic and dynamic physiologic characteristics. It has an appropriate anatomic configuration with a muscular belly proximally and a tendinous insertion distally.⁴⁸ With intrafascicular nerve stimulation, the gracilis can be split into two distal myotendinous units. This may be suitable for providing at least partial independence of finger versus thumb flexion. The gracilis is ideal for transplantation because of its neurovascular structures. It has a dominant pedicle with a single artery and vena comitans and a large, single motor nerve. As a strap muscle it provides adequate strength and excursion for forearm reconstruction.

The gracilis muscle lies in the medial thigh just posterior to the adductor longus. It originates at the pubis and ischium as it courses distally along the inner thigh. It inserts through a strong tendon just posterior to the tibial tubercle on the medial shaft of the tibia. The origin of the adductor muscle is easily palpated in the upper inner thigh. This provides the landmark for the gracilis incision. The incision is made just posterior to the adductor longus, and the muscle interval between the adductor and the gracilis can often be palpated in thin patients (Fig. 54-13, C). This incision should parallel this muscle interval in the upper inner thigh and course distally to the junction of the middle and distal thirds of the thigh. The dissection proceeds deep toward the muscle, which is usually easily identified. The neurovascular pedicle is then approached by

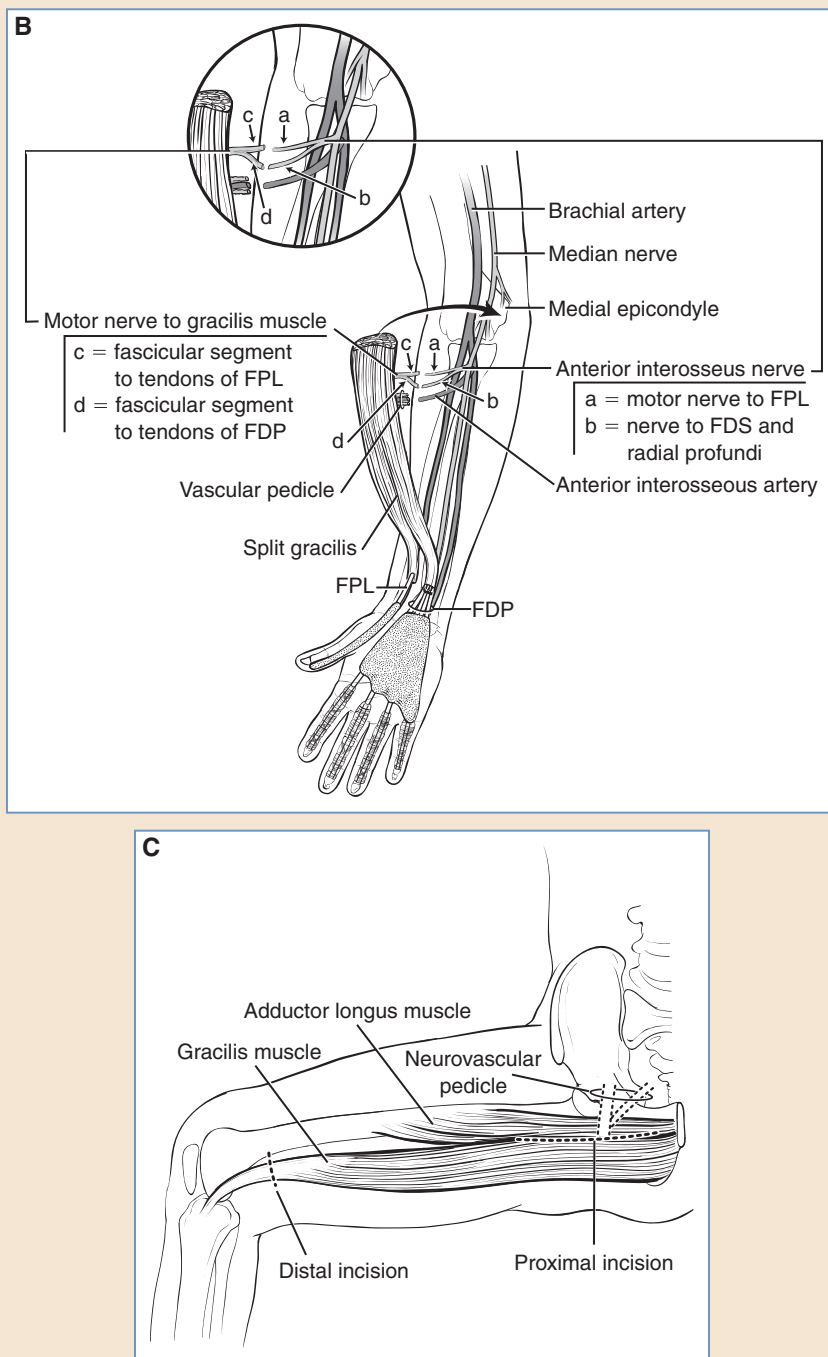


Fig. 54-13, cont'd B, Separation of function for thumb and finger flexion. The inset shows an expanded view of the neurovascular pedicle of the gracilis when the muscle is transferred to the forearm for microvascular anastomosis to the anterior interosseous vessels and microneural coaptation to the anterior interosseous nerve. **C**, Incisions for harvesting the gracilis muscle. (*FDP*, Flexor digitorum profundus; *FDS*, flexor digitorum superficialis; *FPL*, flexor pollicis longus.)

elevating the adductor longus anteriorly. As the adductor longus is elevated, the vascular pedicle comes into view. It generally enters the muscle approximately 8 to 12 cm from its origin and onto the deep surface. This is the major pedicle. It has a dominant artery and two venae comitantes. The motor nerve enters the pedicle just proximal to the vessels at a 45-degree angle. The vascular pedicle is dissected back to easily identifiable branches going upward into the adductor longus. These are divided, and the rest of the pedicle is easily visualized going deeper to its takeoff from the profunda femoris artery with its vena comitans. Very often, the paired venae comitantes unite at the takeoff from the profunda femoris vein.

The entire gracilis is elevated from the thigh except for its origin, tendinous insertion, and neurovascular pedicle. Marking the muscle at this stage to provide tension guides after transplantation is essential. The muscle is placed in a fully stretched position by abducting the hip and extending the knee. Markers are placed at 5 cm intervals with the muscle in this position. This can be done with sutures that will be readily identified after transplantation (Fig. 54-13, *D*).

The motor nerve to the gracilis is a branch of the anterior division of the obturator nerve (L2-3). It takes an oblique course between the adductor longus and adductor magnus. In general, this nerve contains two or three fascicles, and, with intraoperative stimulation, each fascicle can be stimulated and the appropriate motor territory observed. This can be facilitated by palpation of a portion of the muscle with individual fascicle stimulation. The myotendinous units are split distally and separated. They are seen pulled out to length in Fig. 54-13, *E*. With stimulation of one fascicle, one segment of the split muscle contracts (Fig. 54-13, *F*). This separation has potential use for the reconstruction of independent finger and thumb flexion.

After the markings have been placed at 5 cm intervals along the course of the muscle, and the distal tendinous segment of the gracilis is divided if necessary, the origin to the muscle is divided (Fig. 54-13, *G*). This leaves the muscle attached only by its neurovascular pedicle. Only when the forearm dissection is complete and ready to receive the transplant should the neurovascular pedicle be divided. This minimizes muscle ischemia.

The muscle is removed from the thigh and transplanted into the forearm (Fig. 54-13, *H*). The donor site in the thigh is closed in layers. The fascia is closed loosely to reduce the likelihood of muscle herniation. Usually, a suction drain is placed.

The muscle is positioned in the forearm to minimize the distance between the nerve repair and the muscle itself. This will reduce the time required for innervation and assist the return of adequate muscle function. Placing the muscle in the appropriate position for nerve repair allows adequate placement of the muscle belly over the medial epicondyle for the origin and placement of the tendon over the distal forearm for the insertion. The surgeon stretches the muscle to its fully extended position, as indicated by the tension guide sutures. The origin and insertion are visualized and the muscle bellies and/or tendon shortened to facilitate optimal positioning of the muscle.

Repair of the Neurovascular Structures

The muscle origin is then securely fixed to the medial epicondyle, and the neurovascular structures are repaired. Under an operating microscope, the pedicle of the gracilis is prepared. The larger vena comitans is isolated and prepared for microvascular anastomosis. This vena comitans often needs to be separated from the artery to facilitate repair. The microvascular venous repair is carried out first to either a superficial vein in the proximal forearm or the vena comitans of the anterior interosseous artery. Next, the artery to the gracilis is repaired to the anterior interosseous artery. These small vessels require technically perfect anastomoses with every attempt made to prevent thrombosis. Revision is possible but increases the ischemia time and the likelihood that the muscle will be irretrievably damaged. After revascularization is carried out, the entire muscle should appear healthy and revascularized and should contract on direct stimulation. This is often the best guide to muscle viability at this stage of revascularization.

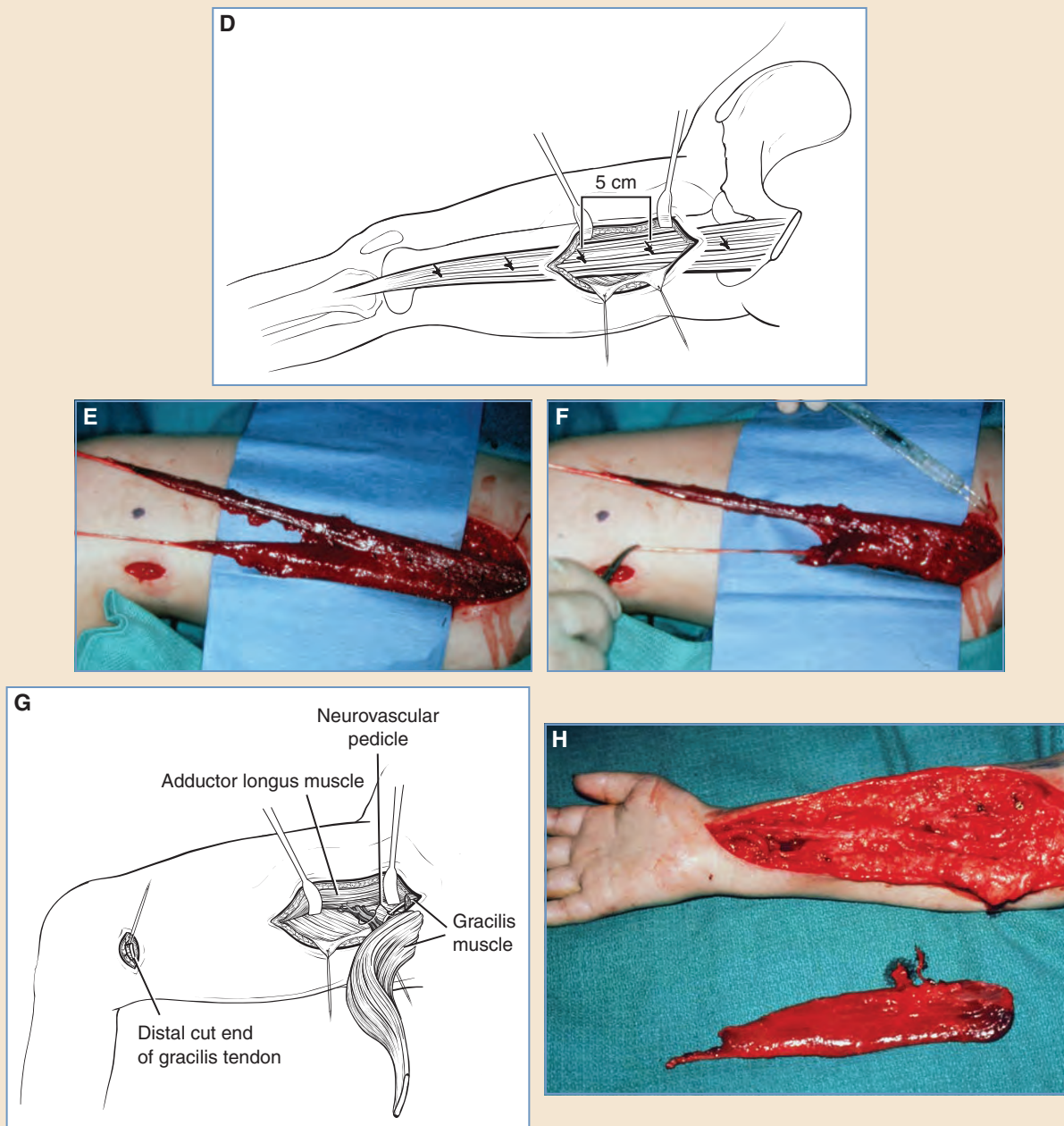


Fig. 54-13, cont'd **D**, Tension guide sutures are placed at 5 cm intervals on the fully stretched muscle. **E** and **F**, Segmental contraction of the gracilis muscle with individual fascicle stimulation. Muscle segments are pulled to length (**E**). A single fascicle is stimulated, selectively contracting one segment of muscle (**F**). **G**, Harvest of the gracilis muscle. **H**, Muscle is removed from the thigh and ready for insertion in the forearm.

The motor nerve is then repaired. This is done as close to the gracilis muscle as possible to minimize reinnervation time. The fascicles are accurately aligned under high magnification and tacked together as perfectly as possible. If a split gracilis is required, then the nerve repairs are evaluated very carefully. The fascicle of the anterior interosseous nerve that innervates the profundus is coapted to the fascicle of the gracilis motor nerve that innervates the section of the gracilis that will flex the finger. Similarly, the fascicle of the anterior interosseous nerve that innervates the flexor pollicis longus is coapted to the fascicle of the gracilis motor nerve that innervates the section of the gracilis that will flex the thumb. These nerve repairs should be perfect, under no tension, and as close to the muscle as possible.

Tendon Repair

Before the tendons are repaired, the muscle is stretched to its fully extended position, as indicated by the previously placed tension guide markers. The wrist and the fingers are also fully extended, and the location of the tendon repair is noted (Fig. 24-13, *I* and *J*). In this way, wrist and finger extension will be possible, and the muscle will flex at its most mechanically advantageous position. The site of tendon repair is marked. The wrist and fingers can be brought into a flexed position and a woven, secure tendon repair carried out without tension. If a split gracilis is required, then the portion of the gracilis tendon that will be used for finger flexion is woven through the balanced profundus. The portion of the gracilis that will be used for thumb flexion is woven through the flexor pollicis longus. The thumb flexor should be in a slightly more lax position relative to the fingers. With contraction, the thumb will not be pulled prematurely into the

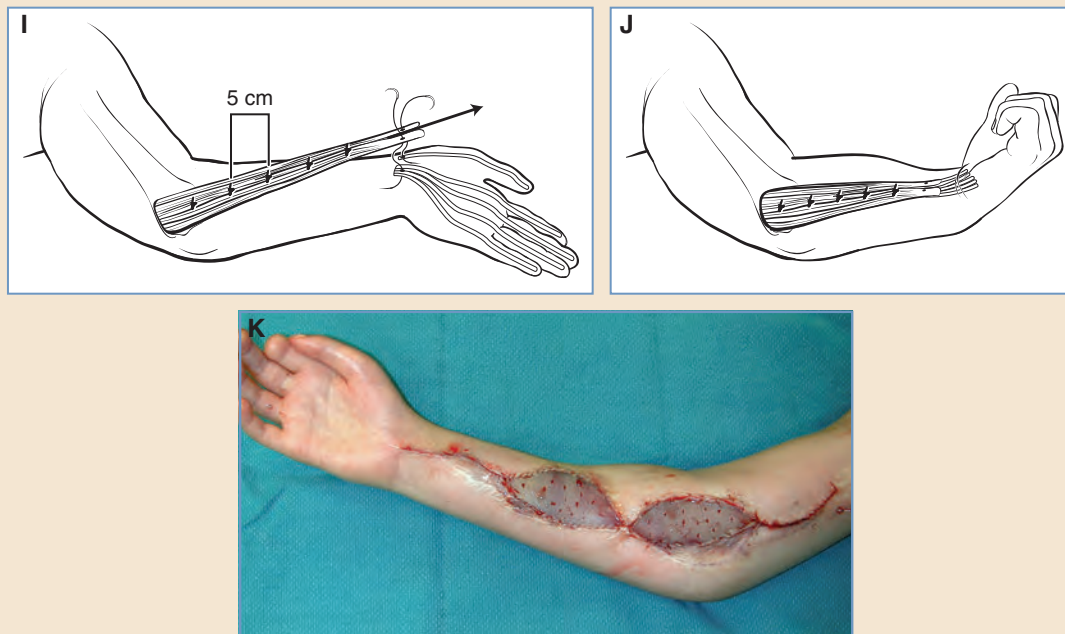


Fig. 54-13, cont'd **I**, The muscle in the forearm is pulled to the length confirmed by tension guide sutures and by the site of the marked tendon repair. **J**, The tendon of the gracilis muscle is repaired by the woven technique to balance the tendons of the forearm at the designated site. **K**, The position of the extremity on completion of the transplant.

palm, which would diminish grip. Placing the muscle under appropriate tension is exceedingly important for optimizing muscle function.

The site of tendon repair is covered with soft tissue. A healthy flap over this site will allow tendon gliding. On the other hand, the muscle belly can be covered with split-thickness skin grafts to prevent excessive bulk that can be caused by flap coverage. The adjacent skin flaps on either side of the incision can be used to cover the crucial areas of the vascular and neural repairs, with the central segment of the muscle belly left open with split-skin coverage. Drains are placed at a distance from the vascular repairs. At the completion of the procedure, the extremity is immobilized. It is preferable to have the elbow at 90 degrees and the wrist slightly flexed to relieve tension from the origin and insertion of the muscle. The finger metacarpophalangeal joints are maintained in a flexed position of 90 degrees, and the thumb is abducted with the interphalangeal joint slightly flexed (Fig. 54-13, K).

Postoperative Care: Childhood Volkmann Ischemic Contracture

As in any free tissue transplant, optimizing circulation during the early postoperative phase is critical. Thus a high-circulating blood volume, with a stable blood pressure and a hematocrit at optimum viscosity for tissue perfusion, is essential. The limb should be placed in a slightly elevated position and the patient gradually mobilized from bed rest to a chair to ambulation over a period of several days.

As indicated previously in this chapter, rehabilitation is essential for the success of the operation. The collaborative efforts of the patient, physical therapist, occupational therapist, and the surgeon are needed. For the first 3 weeks postoperatively, the extremity is splinted in a position that relaxes the tendon repairs distally and the site of muscle belly fixation proximally. This is followed by a 3-week period of gradual passive exercises. Initially, the fingers and wrist are brought into full extension with the elbow flexed. Then the elbow is fully extended with the wrist and fingers fully flexed. Over the next 3-week period, the fingers, wrist, and elbow are simultaneously extended. After this, a second 3-week period of gradual passive exercises is carried out. Initially, the fingers and wrist are brought into full extension with the elbow flexed. The elbow is then fully extended with the wrist and fingers fully flexed. During a third 3-week period, the fingers, wrist, and elbow are simultaneously extended. In this manner, after 9 weeks, the patient has undergone full passive mobilization. At this stage, the beginnings of active muscle contraction will become evident. When active muscle contraction begins, beginning active exercises to gain excursion and minimize adhesions, particularly distally, is critical. As the muscular contraction increases, the digital and thumb excursion should also increase. Active exercises with resistance should be started as soon as it is possible. This will further increase excursion and strength in particular. The entire rehabilitation program may be of ongoing benefit for up to 1½ years, until it plateaus. Both patient and family involvement are essential.

Complications: Childhood Volkmann Ischemic Contracture

Because these procedures are long and complex, particular care should be directed toward patient safety. The children must be well padded intraoperatively and kept warm. Pressure sores are prevented by constant vigilance during the procedure and during the early postoperative phase. Surgical complications can be either early or late. Bleeding can be a problem in the early phases in a densely scarred and highly vascular dissection. Infection can occur during long surgeries with open wounds. Routine intraoperative and postoperative antibiotic coverage is necessary. Wound breakdown and delayed healing can occur in these large wounds. Surgeons should anticipate a degree of delayed wound healing and not let this interfere with the rehabilitation program.

Late complications are more common; of significance are tendon adhesions at the site of tendon repair. In a series of seven patients, Zuker et al³⁰ noted that three tenolyses were necessary.

The likelihood of this complication can be reduced by early passive digital movement, but this may not be very easy in a young child. The second major late complication is wrist flexion deformities. The transplanted forearm flexor is more powerful than the extensors, leading to a fixed flexion deformity. This can be counteracted with splinting during the early phases. However, as growth continues, children undergo growth spurts that can lead to a wrist volar contracture formation. Thus diligence, particularly during growth spurts, with splinting of the wrist in neutral or extension, is extremely important. Patients should be observed and followed until their growth is complete. Even with appropriate diligence and splinting, volar wrist contractures can develop and may require such measures as serial splinting, serial casting, and even surgical intervention to facilitate realignment.

Results: Childhood Volkmann Ischemic Contracture

The results of muscle transplantation in patients with Volkmann ischemic contracture have been extremely rewarding. Muscle contraction usually begins 2 months postoperatively, and improvement is seen up to 1½ years after surgery. The main benefit is finger and thumb flexion, thus improvement in grip strength (Fig. 54-14). Results vary, however, from patient to patient. Many factors account for these differences. First, factors that affect the overall function of the extremity include the severity of the initial injury and particularly the condition of the median and ulnar nerves in the hand. These greatly influence a child's ability to perform fine movements of the hand, which are controlled by the intrinsic musculature. Second, the clinical course after the initial injury can be very influential. Open wounds can become desiccated after a fasciotomy, decompression, and debridement, leading to further necrosis and increased scarring. Infection leads to further tissue destruction and fibrosis. Joints that are allowed to become stiff are considerably more difficult to mobilize. Third, the technical aspects of the muscle transplant itself can influence the results. Perhaps most important are muscle positioning and tension. If the muscle is too tense, the digits will be flexed and even passive extension will be restricted. If it is too lax, the full effect of muscle contraction will not occur. The muscle must be viable; thus technically perfect microvascular repairs and continuation of muscle perfusion must be ensured. The motor nerve that will innervate the transplant needs to be healthy and appropriate for the function desired. The technical microneural coaptation must be perfect. After transplantation, the extremity is properly immobilized to prevent dehiscence and stretching of the tendon repairs. Patients must begin and comply with a graduated, controlled rehabilitation program to gain as much function as possible.

Barring significant complications, complete viability and function of the muscle can be expected. An excellent range of finger flexion can be anticipated, with proper positioning of the



Fig. 54-14 A classic preoperative appearance with the wrist in slight flexion, the metacarpophalangeal joints in extension, the interphalangeal joints in flexion, and the thumb in adduction.



Fig. 54-15 Postoperative photos of a patient who underwent a gracilis muscle transplantation to the forearm to restore finger and thumb flexion. **A**, Full wrist and finger extension with no volar tightness. **B**, Full finger flexion. **C**, A powerful gross grasp. **D**, The bulk of the gracilis muscle is evident on contraction.



Fig. 54-16 A secondary tenolysis is performed to gain greater digital excursion.

muscle. The distance of the finger pulp to the palm should be 3 cm or less.³¹ A perfectly coordinated grip is not a realistic expectation when the muscle transplant is used for both finger and thumb flexion. However, the finger excursion should be within 3 cm of the palm, and the thumb should flex slightly later than the fingers (Fig. 54-15). The thumb should not flex in advance of the fingers and interfere with grip.

When palpable muscle contraction is adequate but digital excursion is not, a tenolysis at the site of the tendon repair may be very useful. This can be anticipated in almost 50% of cases.³¹ This procedure frees the tendon repair from unwanted fibrous adhesions and allows greater excursion (Fig. 54-16). However, great care is required to protect the vulnerable median and ulnar nerves from inadvertent injury. If muscle tension is too lax because of inappropriate positioning or tendon stretching, a tendon-shortening procedure may be helpful.

With a successful transplant, muscle power will approach 50% of normal in an adult but only 25% of normal in a child. This significant difference is probably a reflection of testing techniques and compliance in the rehabilitation program (Fig. 54-17).

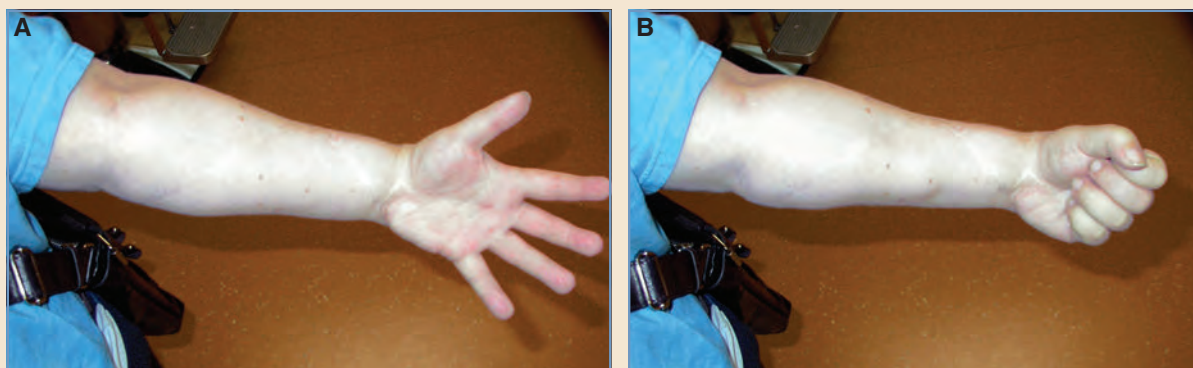


Fig. 54-17 Late results of a gracilis muscle transplantation in a patient with Volkmann ischemic contracture. **A**, Full extension with minimal flexor tightness. **B**, Adequate flexion for an effective grip, with the thumb in an appropriate position.

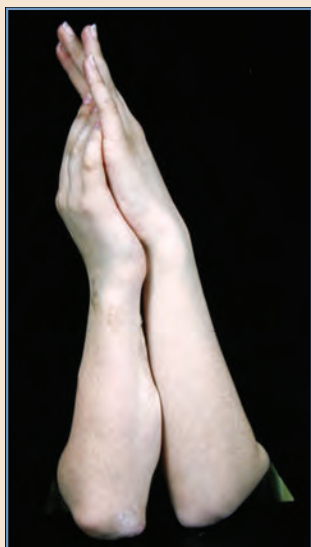


Fig. 54-18 Unresolved problems after Volkmann ischemic contracture in a child can include a limb-length deficiency.

UNRESOLVED PROBLEMS WITH CHILDHOOD VOLKMANN ISCHEMIC CONTRACTURE

Despite the success of muscle transplantation to the forearm for Volkmann ischemic contracture in children, many significant problems remain. Because the growth plates are very vulnerable to ischemic damage, a limb-length discrepancy can occur (Fig. 54-18). This is often compounded by extensive scarring throughout the forearm. There is currently no effective solution to this problem. Limb lengthening, although highly successful in other situations, often leads to an unacceptable functional loss in the forearm in an already compromised limb.

A second problem involves the loss of pronation and supination. Extensive scarring along the length of the forearm bones restricts movement and can significantly compromise the positioning of the hand. Even with major releases and insertion of interposition materials, major limitations persist.

Last, the delicate balance of the musculature of the forearm is often irreparably damaged. The transplanted muscle on the volar surface may overpower the weakened extensors. With time, and particularly with growth spurts, the wrist may be pulled into flexion. The persistent pressure on the wrist can lead to a significant bony malpositioning of the carpal bones. Therefore surgeons should anticipate problems with wrist flexion throughout the remaining growth of the forearm bones, especially during the growth spurts. Night splinting and even serial casting may be required to minimize this potentially devastating complication.

KEY POINTS

Neonatal Volkmann Ischemic Contracture

- Proper management requires early diagnosis and treatment.
- The less severe injuries involve the dorsal muscle groups.
- Early fasciotomy on both the dorsal and the volar compartments is necessary; volar compartment injuries are often not recognized.
- All necrotic muscle is removed, but muscles with any degree of function are not debrided.
- Neurolysis of the median, ulnar, and radial nerves is worthwhile.
- Newborns' wounds are skin grafted, but full-thickness coverage is provided before multiple, complex, secondary reconstructive procedures are performed.
- Skeletal growth will be abnormal in all but type I injuries.

Childhood Volkmann Ischemic Contracture

- Distraction lengthening should not be performed.
- Prevention is the best treatment.
- Deep volar components are the most severely affected.
- The process can affect volar and extensor surfaces.
- Angiograms are useful to not only define vasculature but also to indicate the condition of the anterior interosseous nerve.
- Stage-setting procedures will optimize outcomes.
- Muscle transplantation can be effective to provide flexion, extension, or both.
- Providing digital balance and appropriate muscle tension during a functioning muscle transplantation is crucial.
- A powerful, healthy motor nerve should be selected.
- Compliance with splinting and a concentrated rehabilitation program is essential, emphasizing the importance of patient selection.

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Pediatric Soft Tissue and Bony Sarcomata of the Extremities: Considerations and Reconstruction

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arcomas are a heterogeneous group of tumors of mesenchymal origin. Osteosarcoma, Ewing sarcoma, and rhabdomyosarcoma are the most common pediatric sarcomas, accounting for 14% of pediatric malignancies.¹ Although the prognosis for metastatic disease remains poor with 5-year survival rates of less than 30%,²⁻³ 5-year survival rates for localized disease have improved over the last 30 years, from 10% to 70% in osteosarcoma² and from less than 10% to 50% in Ewing sarcoma.³ In rhabdomyosarcoma, 5-year survival is as high as 94%.⁴

The management of pediatric sarcomas requires a multidisciplinary approach, including a wide range of surgical expertise, depending on the site of presentation, medical oncology, radiation oncology, radiology, pathology, rehabilitation therapy, nursing, and psychosocial support. As improvements in management have increased overall survival, quality of life has become a more important consideration in the treatment of pediatric sarcoma. The aims of this chapter are to provide a brief overview of the surgical approach to pediatric sarcoma and to describe the reconstruction of sarcoma defects, particularly for tumors of the extremities.

Osteosarcoma is an aggressive primary bone tumor that commonly presents in the long bones. Most cases occur in the absence of inherited syndromes or exposure to radiation. Associations with syndromes such as hereditary bilateral retinoblastoma, Li-Fraumeni syndrome, Bloom syndrome, Rothmund-Thomson syndrome, multiple exostoses, and Paget disease have been described.² The peak incidence of osteosarcoma is at 16 years of age, or during puberty, suggesting an association

with a high rate of bone growth. Approximately 20% of patients present with metastases, which are most frequently found in the lung.⁵ Osteosarcoma appears as both osteoblastic and osteolytic on plain radiographs, with an aggressive periosteal reaction; a triangle of immature bone at the edge of the bone cortex is described as a *Codman sign*. Skip metastases are found infrequently and should be identifiable on bone scan, CT, and MRI. The introduction of chemotherapy in conjunction with surgery in the 1970s as a treatment for presumed micrometastases led to the significant improvement in survival noted previously. Osteosarcoma is fairly resistant to radiotherapy.

Ewing sarcoma is part of a family of tumors that also includes primitive neuroectodermal tumors. The peak incidence is in the second decade of life.³ Although most Ewing sarcoma is found in bones, particularly in diaphyses, metaphyses, and the pelvis, soft tissue tumors have also been described. Approximately 25% of patients present with metastases most often found in the lungs, bone, and bone marrow.⁶ On plain radiographs, Ewing sarcoma has an onion-skin periosteal reaction and a moth-eaten appearance to the cortex. Soft tissue extension of the tumor often occurs early. Surgery, chemotherapy, and radiotherapy are all employed in the treatment of patients with Ewing sarcoma.

Rhabdomyosarcoma is the most common soft tissue sarcoma in children, and nearly two thirds of patients are younger than 10 years of age at presentation.⁴ The peak incidence is bimodal, occurring in children 2 to 6 years of age and 15 to 19 years of age.¹ Rhabdomyosarcoma can develop in any soft tissues of the body and is grouped according to anatomic sites; the genitourinary tract and the extremities are the most common locations. Fifteen to twenty percent of patients present with metastases that can be widespread.⁷ Surgery, chemotherapy, and radiotherapy are useful modalities in the management of rhabdomyosarcoma.

The other soft tissue sarcomas are grouped collectively and known as *nonrhabdomyosarcoma*. They are generally managed by surgical excision, and the use of radiotherapy and chemotherapy is inconsistent. The most common soft tissue sarcoma is *synovial sarcoma*, which is a rare tumor. It presents most often as a painless mass in the upper or lower extremities and has been associated with the chromosome translocation t(x;18)(p11;q11), in which segments of chromosome 18 and chromosome x have switched positions.

STAGING OF SARCOMAS

Two different staging systems are used for sarcomas.⁸ The Musculoskeletal Tumor Society, or the Enneking system, classifies benign musculoskeletal tumors as latent, active, or aggressive⁸ (Table 55-1). Malignant tumors are classified based on grade (low or high), whether the tumor is intra-compartmental or extracompartmental, and the presence of metastasis⁹ (Table 55-2). This system is useful for guiding surgical resection, because anatomic features of the tumors are included.

The American Joint Committee on Cancer (AJCC)¹⁰ employs the TNMG staging system for bone cancers. It is based on the size and depth of the primary tumor, the histologic grade, the regional lymph node status, and the presence of metastasis (Box 55-1). The Intergroup Rhabdomyo-

Table 55-1 Enneking Staging for Benign Musculoskeletal Tumors Based on Radiographic Characteristics of the Tumor Host Margin

Stage	Description
Latent	Well-demarcated borders
Active	Indistinct borders
Aggressive	Indistinct borders

Table 55-2 Enneking Staging for Malignant Musculoskeletal Tumors Based on Surgical Grade, Local Extent, and Presence of Metastasis

Stage	Grade	Site	Metastasis
IA	Low (G1)	Intracompartmental (T1)	No metastasis (M0)
IB	Low (G1)	Extracompartmental (T2)	No metastasis (M0)
IIA	High (G2)	Intracompartmental (T1)	No metastasis (M0)
IIB	High (G2)	Extracompartmental (T2)	No metastasis (M0)
III	Any (G)	Any (T)	Regional or distant metastasis (M1)

Box 55-1 American Joint Committee on Cancer Staging System**T Stages of Bone Cancer**

TX: The primary tumor cannot be measured.

T0: No evidence of the tumor

T1: The tumor is 8 cm (about 3 inches) or less.

T2: The tumor is larger than 8 cm.

T3: The tumor is in more than one place on the same bone.

N Stages of Bone Cancer

N0: The cancer has not spread to the lymph nodes near the tumor.

N1: The cancer has spread to nearby lymph nodes.

M Stages of Bone Cancer

M0: The cancer has not spread anywhere outside of the bone or nearby lymph nodes.

M1: Distant metastasis (The cancer has spread.)

– M1a: The cancer has spread only to the lung.

– M1b: The cancer has spread to other sites (such as the brain and/or the liver).

Grades of Bone Cancer

G1-G2: Low grade

G3-G4: High grade

Stage Grouping

- Stage I: All stage I tumors are low grade and have not yet spread outside of the bone.
 - Stage IA: T1, N0, M0, G1-G2: The tumor is 8 cm or less.
 - Stage IB: T2 or T3, N0, M0, G1-G2: The tumor is either larger than 8 cm or it is in more than one place on the same bone.
- Stage II: Stage II tumors have not spread outside the bone (like stage I) but are high grade.
 - Stage IIA: T1, N0, M0, G3-G4: The tumor is 8 cm or less.
 - Stage IIB: T2, N0, M0, G3-G4: The tumor is larger than 8 cm.
- Stage III: T3, N0, M0, G3-G4: Stage III tumors have not spread outside the bone but are in more than one place on the same bone. They are high grade.
- Stage IV: Stage IV tumors have spread outside of the bone they started in. They can be any grade.
 - Stage IVA: Any T, N0, M1a, G1-G4: The tumor has spread to the lung.
 - Stage IVB: Any T, N1, any M, G1-G4 or any T, any N, M1b, G1-G4: The tumor has spread to nearby lymph nodes or to distant sites other than the lung (or both).

T = Features of the tumor (its size and whether it is in more than one spot on the bone); N = spread to lymph nodes; M = metastasis (spread) to distant organs; G = tumor grade.

Table 55-3 Intergroup Rhabdomyosarcoma Study Group Site-Modified Tumor, Metastasis, Number Staging

Stage	Sites	T	Size	N	M
1	Favorable	T1 or T2	a or b	N0 or N1 or Nx	M0
2	Unfavorable	T1 or T2	a	N0 or Nx	M0
3	Unfavorable	T1 or T2	a b	N1 N0 or N1 or Nx	M0 M0
4	All	T1 or T2	a or b	N0 or N1	M1

Definitions: Favorable sites = Orbit, head, and neck (excluding parameningeal), genitourinary (excluding bladder/prostate tumor), and biliary tract; unfavorable sites = bladder/prostate, extremity, cranial parameningeal, and other sites (including the trunk and retroperitoneum, but excluding biliary); T1 = confined to the anatomic site of origin, T2 = extension and/or fixation to the surrounding tissue; size a ≤ 5 cm in diameter, b > 5 cm in diameter; regional nodes N0 = regional nodes not clinically involved, N1 = regional nodes clinically involved by the neoplasm, Nx = clinical status of regional nodes unknown (especially sites that preclude lymph node evaluation); metastasis M0 = no distant metastasis, M1 = metastasis present. The pretreatment size is determined by external measurement using MRI or CT depending on the anatomic location. For less accessible primary sites, CT will be employed as a means of lymph node assessment. Metastatic sites will require some form of imaging but not histologic confirmation, except for bone marrow examination.

sarcoma Study Group modified an AJCC TNM system for soft tissue sarcomas to classify tumors based on site (favorable or unfavorable), whether the tumor is confined to the anatomic site of origin or extends into surrounding tissue, whether the tumor is greater than or less than 5 cm in diameter, the regional lymph node status, and the presence or absence of metastasis⁴ (Table 55-3).

DIAGNOSIS AND STAGING INVESTIGATIONS

Sarcomas present in a variable manner depending on the tissue of origin. Bone tumors often present with pain, swelling, and potentially a pathologic fracture, and soft tissue sarcomas typically present with swelling and a firm mass. Imaging is a key component of diagnosis and should take place before a surgical biopsy. Modalities, including radiography, MRI, and CT, help to define the tumor characteristics. For soft tissue lesions, ultrasound can be useful as a primary modality in younger patients in whom general anaesthesia may be required for MRI and judicious use of CT scans should be employed. Once the local imaging is complete, systemic imaging should be performed and may include further MRI to examine lymph node basins, CT scans of the chest and abdomen, whole-body bone scans, PET scans, and possibly bone marrow aspiration and biopsy for Ewing sarcoma and rhabdomyosarcoma. Bloodwork includes a CBC; an erythrocyte sedimentation rate (ESR) and C-reactive protein (CRP), both of which may be elevated with Ewing sarcoma, infectious causes, and lymphoma; and electrolytes, including calcium, magnesium, and phosphate. Serum alkaline phosphatase has been shown to correlate with 5-year, event-free survival in osteosarcoma.¹¹ Similarly, serum lactate dehydrogenase has prognostic value in osteosarcoma¹² and Ewing sarcoma.¹³

A tumor biopsy should be obtained with careful consideration of future requirements for surgical excision. The biopsy tract must be excised at the time of tumor resection. For tumors on the extremities, biopsies should be longitudinal along an extensile incision whenever possible (Fig. 55-1). Sampling multiple biopsy sites is not recommended. When these principles are not followed, the soft tissue reconstruction of the resulting defect may become unnecessarily complicated.



Fig. 55-1 A longitudinal biopsy incision line along an extensile incision.



Fig. 55-2 Low-grade infantile fibrosarcoma of the upper extremity.

SURGICAL MANAGEMENT

The primary guiding principle in the surgical management of pediatric sarcoma is oncologic safety. Complete resection of the tumor is factored into the staging and may indicate further treatment depending on the type of tumor. Secondary guiding principles include function, durability, complications, and appearance.

Four main types of excision exist for sarcomas⁹:

1. Intralesional excision
2. Marginal excision: The margins of excision are at the margin of the tumor.
3. Wide excision: The margins include a cuff of normal tissue around the tumor.
4. Radical excision: The entire compartment containing the tumor is excised.

The type of excision, or the surgical margin, required is determined by the characteristics of the tumor. For example, in a low-grade lesion such as an infantile fibrosarcoma in the upper extremity (Fig. 55-2), a marginal excision may be performed to maintain function of the limb. A clinical practice guideline developed for the surgical management of soft tissue sarcomas of the extremities recommends that the aim should be to achieve a clear margin, but in certain cases a close or microscopically positive margin may result.¹⁴ In such cases, radiotherapy may be employed as an adjuvant therapy. In high-grade osteosarcoma, the presence of a positive margin has been found to be the only predictor of local recurrence.¹⁵ In these patients, achieving a negative margin should be the aim of surgical management.

Neoadjuvant chemotherapy is commonly employed because of the theoretical increase in safety and the ease of obtaining a negative surgical margin. This rationale holds true for tumors that do not generate a matrix, such as Ewing sarcoma, because they predictably shrink with chemotherapy. However, this rationale does not necessarily apply for osteosarcoma and has not been clinically proven. In one study, surgical resection planning based on MRI changed after the neoadjuvant chemotherapy. The volume of the planned resection changed after chemotherapy but in an inconsistent manner. Some cases required less resection, and others required more than had originally been planned.¹⁶ Neoadjuvant chemotherapy targets potential systematic disease up front. Despite the surgical benefit of neoadjuvant chemotherapy, the known risk of increased wound-healing complications warrants further study.

Surgical Approach

Several key factors should be considered when planning a sarcoma excision:

- The most important factors are to ensure adequate margins based on the type of sarcoma and to determine which key structures can be preserved and which ones cannot.
- Joint involvement. If the bone on one side of a joint cannot be preserved, can the supporting structures (such as the joint capsule and ligamentous support) be preserved?
- Is the outlook for function reasonable, perhaps not normal but better than that offered by an amputation and prosthesis?
- The patient's and family's (for young children) preferences need to be considered.
- Can the joint be safely preserved but at the cost of joint stiffness? Rotationplasty may provide excellent knee function and convert an above-knee amputation to be a below-knee amputation.
- Is an unusual aesthetic appearance acceptable? As in all complex surgical procedures, the options must be carefully discussed with patients and their family.

Five main approaches are available for surgical reconstruction after excision of sarcomas of the extremities (Box 55-2). The first approach, soft tissue reconstruction only, is an option when the defect requires soft tissue cover only. The second, intercalary bony reconstruction, is applicable when an intercalary bony defect results from an excision that preserves the epiphysis. The third, joint resection and reconstruction, requires removal of a joint surface and reconstruction with bone (allograft and/or autograft, that is, fibular head) or with an endoprosthesis. The fourth approach is a rotationplasty in which the knee bony and supporting structures need to be removed. The knee joint is replaced by the ankle joint as the leg is shortened and rotated 180 degrees. It is an almost complete intercalary joint resection combined with a joint replacement. The "new knee" is powered by either the muscles of the calf (after a distal femoral tumor removal) or the muscles of the thigh (after a proximal tibial tumor removal). The fifth surgical approach is an amputation. Historically, this was the most common surgical procedure performed. However, currently, a limb-salvage surgery such as those listed in Box 55-2 is performed in nearly 90% of cases.¹⁷

Soft Tissue Cover Only

When only soft tissue cover is required after a tumor excision, the reconstructive options are numerous. The reconstructive ladder can be used fully. Closure can range from simple, direct approximation of adjacent, clear skin margins to free flap coverage of exposed bone (Fig. 55-3) and/or critical structures. In the patient shown in Fig. 55-3, about 50% of the cortex of the tibia had to be removed. A free latissimus dorsi muscle flap covered this readily, and an overlying split-thickness skin graft provided excellent contour.

Box 55-2 Approaches for Surgical Reconstruction After Tumor Excision of the Extremities

- I. Soft tissue reconstruction only
- II. Intercalary bony reconstruction
 - A. Bone only
 1. Allograft (cadaver bone)
 2. Autograft (fibula)
 3. Allograft and autograft combination
 - B. Bone and soft tissue
 1. Allograft and vascularized coverage
 2. Autograft and vascularized coverage
- III. Joint resection and reconstruction
 - A. Reconstruction with bone
 1. Allograft (cadaver joint)
 2. Autograft (vascularized head of fibula)
 - B. Reconstruction with an endoprosthesis
- IV. Rotationplasty
 - A. New knee powered by calf muscles (distal femoral tumors)
 - B. New knee powered by thigh muscles (proximal tibial tumors)
- V. Amputation



Fig. 55-3 Reconstruction with soft tissue only. **A**, A preoperative view of an extensive tumor initially thought to be a vascular malformation with expanded growth. It was diagnosed as a dermatofibrosarcoma based on a biopsy sample. **B**, The tumor excision involved about 50% of the circumference of the tibia.

Continued

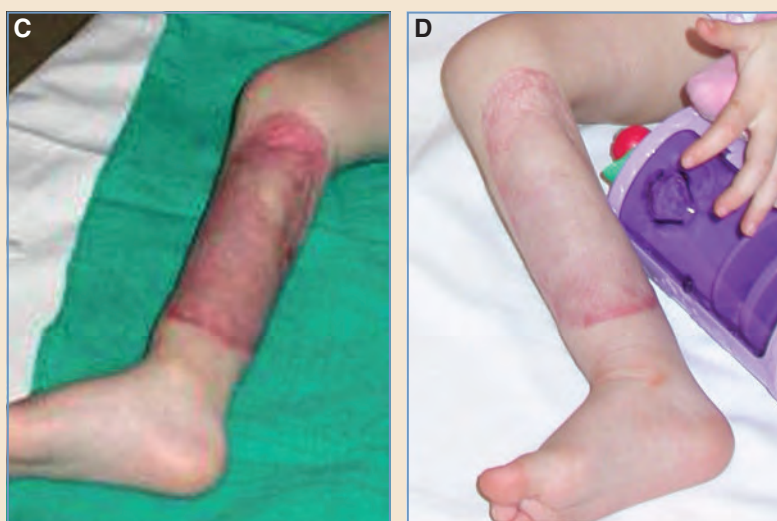


Fig. 55-3, cont'd **C**, A postoperative view after reconstruction with a latissimus dorsi flap and a split-thickness skin graft. **D**, The patient is shown in a late follow-up.

Intercalary Excision and Reconstruction

An intercalary excision is indicated when an MRI shows the adjacent epiphysis is free of tumor and edema. Most children's high-grade bone tumors arise in the metadiaphysis and extend to the epiphysis over time. Although the physis is a relative barrier, it is only a matter of time until it is breached. If, after neoadjuvant chemotherapy, a preoperative MRI scan shows no epiphyseal edema, surgeons can be reassured that no microscopic tumor is present.¹⁸ An osteotomy can be performed on the epiphyseal side of the physis or directly through the physis if preoperative gradual epiphyseal lysis has been undertaken over 2 weeks with a frame (Fig. 55-4). Indications for this procedure are shown in Fig. 55-5.¹⁸

The reconstruction of an intercalary defect has historically included the use of allograft bone. With the development of microsurgical techniques, free vascularized fibula flaps have been incorporated into intercalary reconstruction with improved outcomes. The fibula provides a long segment of bone and possibly a cutaneous skin paddle if required. The intercalary defect can then be reconstructed with bone only or with bone and soft tissue. In a series comparing allograft alone and allograft with an internal vascularized fibula flap, patients receiving a vascularized fibula progressed to weight bearing faster and had fewer revisions but had a similar overall rate of complications.¹⁹ The fibula has been used as a single strut as well as a double-barreled construct. In a series of lower limb reconstruction after sarcoma resection in pediatric patients, the flaps united an average of 4 months after surgery, with patients able to bear weight an average of 6 months after surgery.²⁰ Hypertrophy was a continuing process, with greater than 30% hypertrophy occurring during the first 10 months in all patients, and a range of 30% to 200% hypertrophy after a mean of 38 months. A vascularized fibula flap was also used to salvage allograft nonunion, with an average of 10 months to bony union and 13 to 45 months to unrestricted activity of the limb.²¹ In allograft reconstruction alone, infection rates were reported to be 24% to 30%,²² whereas infection rates with a vascularized fibula flap were reported to be 10% to 14%.^{19,23-25} Reconstruction with a vascularized fibula flap can lead to excellent functional outcomes. In a series of pediatric patients with lower limb sarcoma, 56% returned to ambulation without an aid, and 39% were



Fig. 55-4 A distraction frame is in place to preserve the epiphysis.

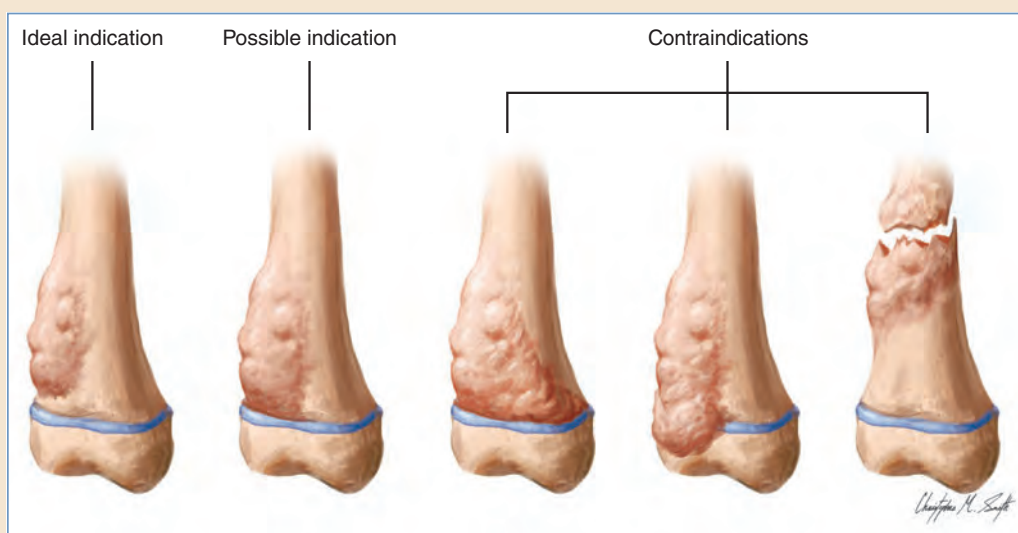


Fig. 55-5 Indications for epiphyseal preservation in metaphyseal malignant bone tumors in children.

able to ambulate with an aid.²⁶ All children of school age were able to return to school, and 50% of children resumed sports.

Technical Considerations Regarding the Vascularized Fibula Flap A vascularized fibula flap is typically planned for the contralateral limb in lower extremity reconstruction to facilitate a two-team approach. A pedicled fibula flap has been used to reconstruct tibial defects that are positioned optimally; however, the inset of the bone can be difficult because of the restricted movement of the pedicle. The patient is positioned on two layers of sponge cushioning to minimize the potential for pressure-induced wounds. Venous thrombosis is rare in children, even in

oncology patients; thus anticoagulation is not employed. Separate operative setups are used to ensure oncologic safety with no contamination of the donor site with tumor, and full change of the surgical instruments is essential once the tumor is excised.

A fibula flap is dissected with a sterile tourniquet on the thigh. A lateral approach to the fibula is used for harvesting an osseous fibula flap, and a posterior approach is used for an osteocutaneous fibula flap, as described in the literature.²⁷ If no skin paddle is required, a longitudinal incision along the posterior border of the fibula is marked, leaving 6 cm of bone proximally and distally in adolescents and as little as 4 cm in younger children. Rarely, a longer length of bone is required, and a syndesmotic screw is placed between the distal fibula and the tibia to ensure joint stability. The bone to be used in the flap is oriented distally to allow maximal length of the pedicle. Even if the amount of bone required for the reconstruction is less than the available length, removing the proximal bone facilitates the dissection of the full length of the pedicle. If a skin paddle is required, the skin is oriented on the distal aspect of the available fibula, and a Doppler probe is used to identify perforating vessels (Fig. 55-6).

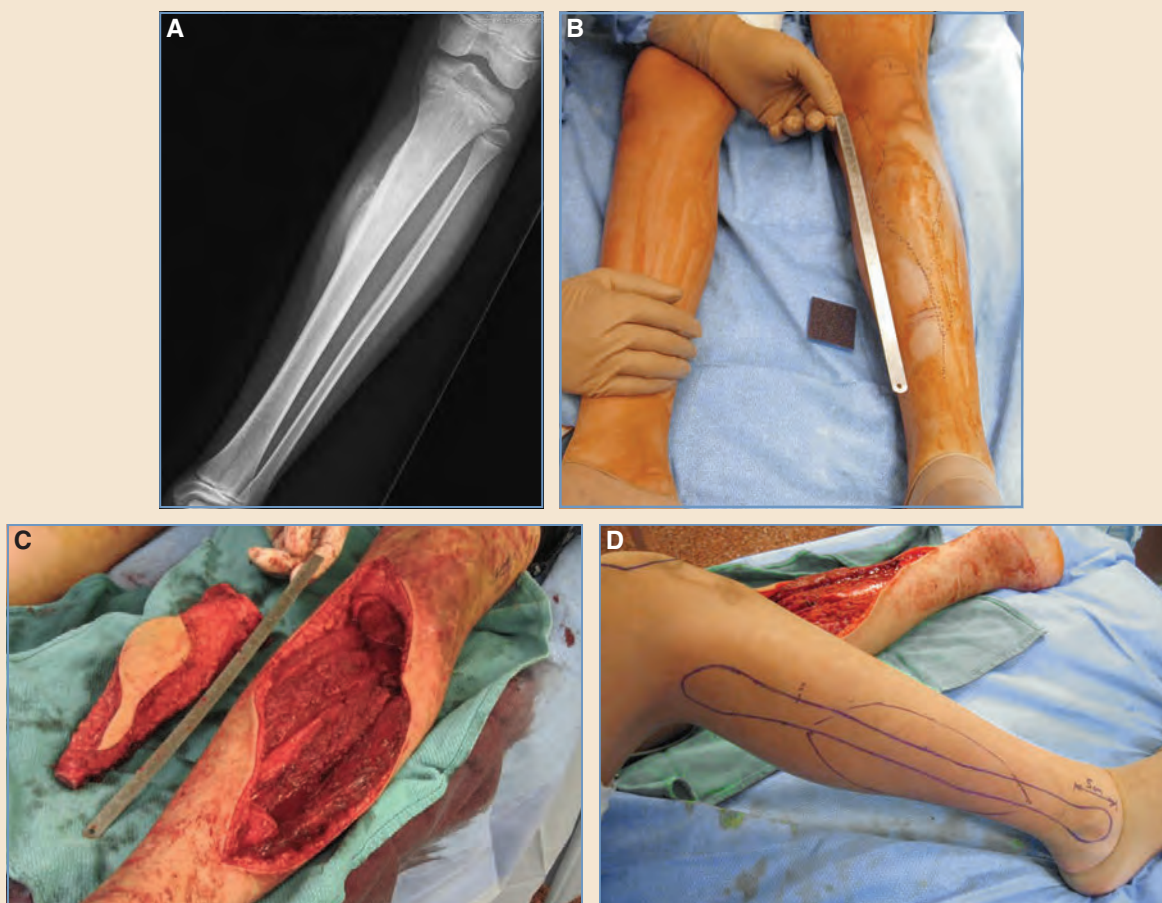


Fig. 55-6 This case shows the role of an osteocutaneous fibula flap for reconstruction after excision of an extensive tumor of the tibia. **A**, A reoperative radiograph of a tumor amenable to intercalary resection. **B**, Intraoperative planning involved measuring the length of fibula required. **C**, The tumor was excised, and the defect was extensive. **D**, An osteocutaneous fibula flap was marked.



Fig. 55-6, cont'd **E**, The flap was harvested. The vascular pattern to the skin component is shown. **F**, Surgical transfer of the flap was complete. Suction drains were placed in the donor site and the recipient site. **G**, Postoperative venous congestion led to loss of most of the cutaneous portion of the flap despite the use of leech therapy. **H**, The patient is shown postoperatively after interval healing with skin grafting of debrided necrotic skin of the cutaneous component of the osteocutaneous flap. **I**, A postoperative radiograph of the reconstructed tibia with vascularized fibula. Solid bony union of the fibula is evident proximally and distally.

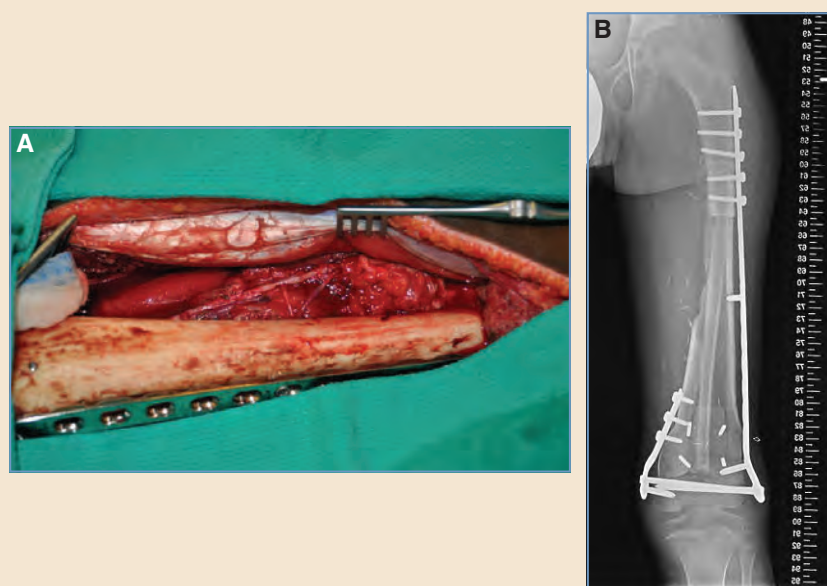


Fig. 55-7 A vascularized fibula provides rapid, solid union, whereas an allograft provides immediate strength. **A**, Intraoperative view of a composite reconstruction with allograft and vascularized autograft. Allograft is stabilized with a plate, and the fibula is underneath it, out of view. A microscope is in place for repairing the peroneal vessels. **B**, A postoperative radiograph of a femoral reconstruction using a large allograft fixed by plates and screws for strength and a vascularized fibula for rapid healing.

The fibula is inset with the orthopedic oncologist present. If the remaining bone is of adequate length, intussusception of the fibula into the remaining bone aids in preventing nonunion.

In patients in whom an allograft is also required to provide early support, a composite reconstruction may be helpful, as popularized by Ceruso et al.²⁸ The fibula is inset in a generous, wide posteromedial slot created in the allograft. It is fixated with long plates but with a minimal number of screws, without violating either the allograft or the autograft, to create a stable construct (Fig. 55-7).

Recipient vessels are identified during the tumor resection. End-to-end anastomoses are preferable, if possible; however, end-to-side anastomoses are often used in wide resections in which few vessels of adequate caliber remain in the operative field.

Postoperatively, patients are placed on a caffeine-free diet. A soft, bulky dressing, occasionally with a splint depending on the proximity of the dissection to a joint, is placed on the reconstructed extremity with adequate visualization of the distal digits to monitor vascularity. With the known high rate of complications with sarcoma surgery, patients are monitored closely in the hospital during their average hospital stay of 15 days.²⁶ Progressing to healing as quickly as possible is important to resume chemotherapy, typically about 2 weeks postoperatively (Fig. 55-8).

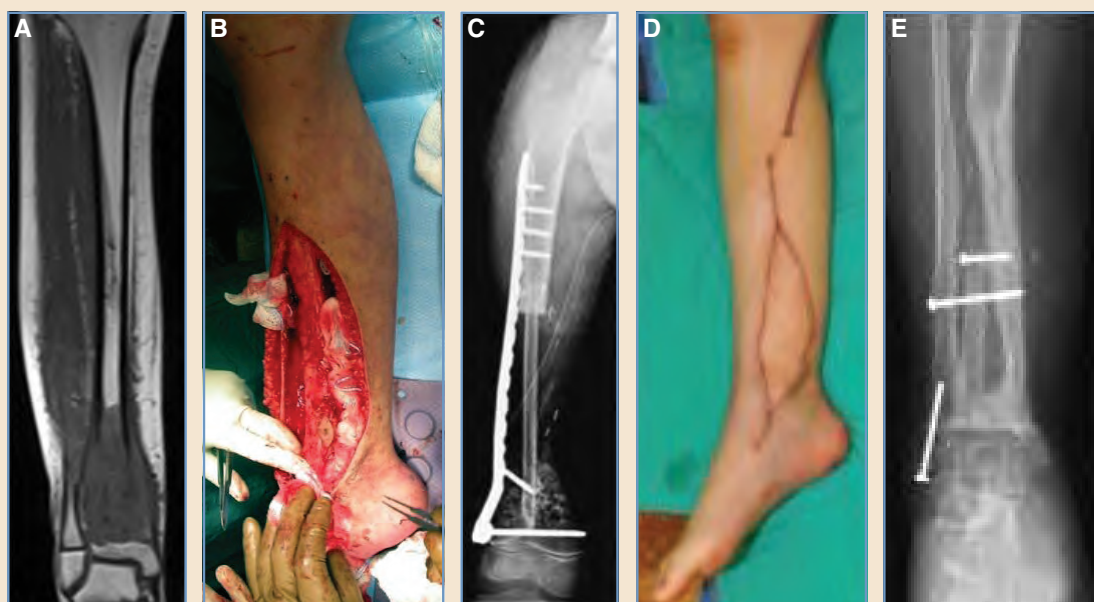


Fig. 55-8 This patient had malignant bone tumors amenable to intercalary resection and reconstruction with a vascularized fibula flap. **A**, Preoperative MRI showed a tumor of the distal tibia. **B**, An intraoperative view after the tumor was dissected free and ready for removal. **C**, A postoperative radiograph showed the transferred contralateral fibula in place. **D**, An intraoperative view at the completion of the osteocutaneous fibula flap transfer. **E**, A radiograph obtained 2 years after the transfer shows excellent bony healing proximally and distally and interval hypertrophy of the transferred fibula.

Joint Resection and Reconstruction

When tumor has involved the epiphysis, most surgeons would resect at least that side of the joint (intracapsular resection). In this situation, when the extracapsular structures can be preserved, a joint reconstruction using bone can be of considerable benefit. Cadaver allograft joint reconstruction has not been reliable or durable. However, the fibula with its head can be transferred as living bone and living joint by microvascular techniques. Thus, for example in young children, resection of the femoral or humeral head can be reconstructed with an osteoarticular fibular free flap (Fig. 55-9). The revascularization has been described based on the peroneal vessels, as discussed earlier in the chapter (see Fig. 55-7), or based on the anterior tibial vessels.²⁹ Over time the fibular head will remodel to some degree to mimic the joint surface of the reconstructed joint. In the patient shown in Fig. 55-10, the ankle mortise was reconstructed with the fibular head, preserving function. This form of reconstruction is not commonly feasible because of oncologic considerations. Oncologic safety remains the priority. If a joint is devascularized as a result of tumor resection, salvage is not possible, and future growth and limb-length discrepancies should be considered when planning the reconstruction. In growing children, this anticipated growth discrepancy can be considerable and functionally limiting, particularly in the lower extremity. They may in time require surgical intervention as opposed to a simple shoe raise. Such surgical options include contralateral epiphyseodesis, acute contralateral shortening, and ipsilateral leg lengthening.

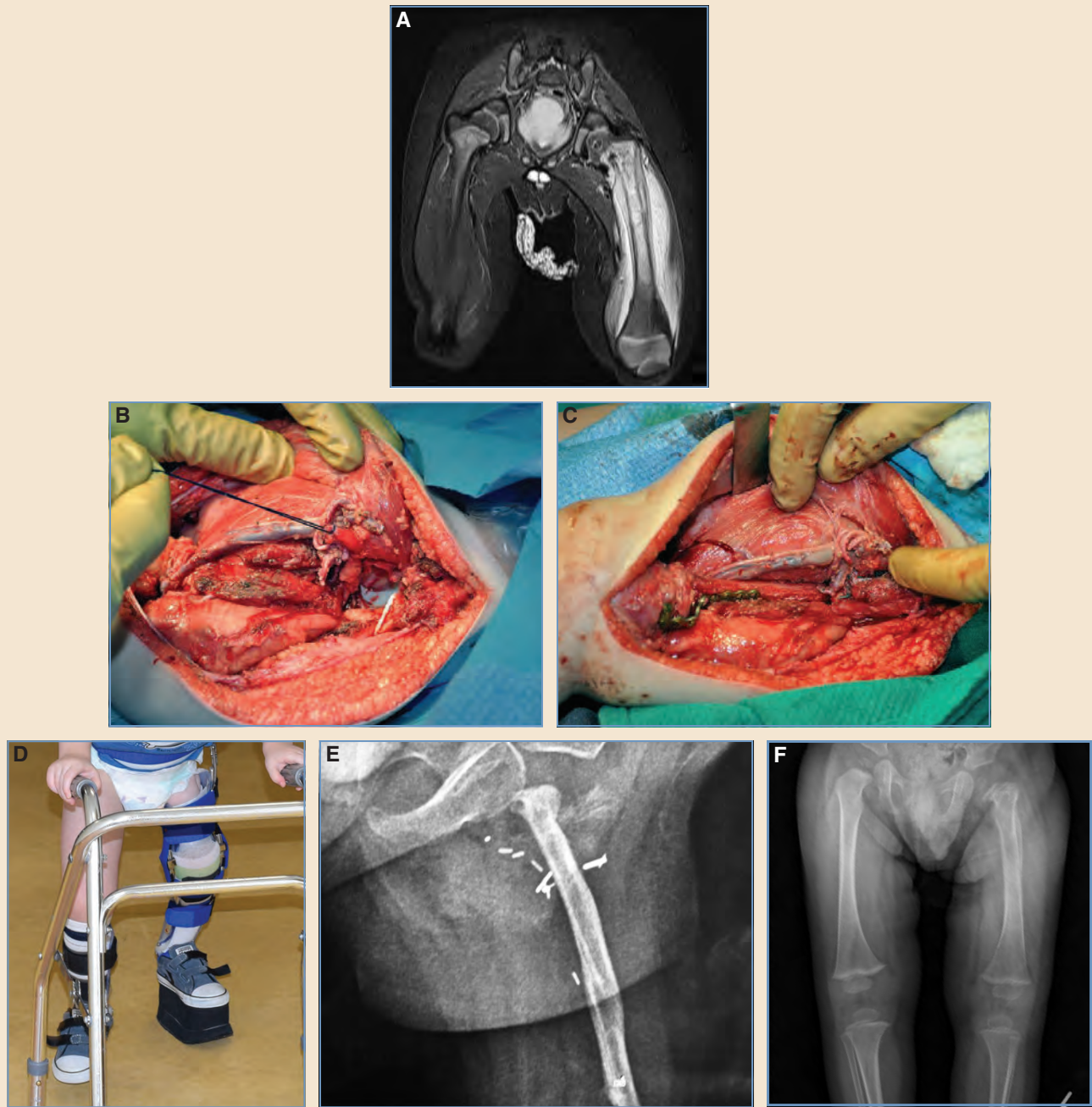


Fig. 55-9 This 1-year-old child favored his left hip during attempted walking. The imaging workup demonstrated a Ewing sarcoma of the femoral head. This was confirmed by an open biopsy specimen. The tumor and femoral head were resected and the femur reconstructed with a free vascularized fibula, including the fibular head. **A**, A preoperative MRI showed the bone tumor of the left femur. **B**, The bony defect after tumor excision. The acetabulum was exposed. **C**, The fibula was positioned, with soft tissue reconstruction proximally and plate and screw fixation distally. **D-F**, The patient is shown 2 years after surgery, chemotherapy, and a rehabilitation program. He is weight bearing and pain free with adequate mobility. Radiographs show hypertrophy of the femur and remodeling of the new femoral head.



Fig. 55-10 This patient underwent a microvascular transfer of the fibular metaphysis, growth plate, and epiphysis for reconstruction of the distal fibula at the distal tibia-fibula joint after resection for osteogenic sarcoma. **A**, The appearance of the leg before tumor excision. The biopsy incision was marked for excision. **B**, A plain radiograph showed an osteogenic sarcoma of the distal fibula. **C**, An intraoperative view of the defect of fibula after the tumor excision. The open joint was without the distal fibula. **D**, Reconstruction of the distal fibula. The proximal fibula with its growth plate and epiphysis was removed, reversed, and revascularized with bone and the peroneal and lateral geniculate vessels. **E**, A postoperative radiograph showed remodeling of the fibular head to create a stable ankle mortise. **F-H**, In a late follow-up, the patient has satisfactory contour and normal dorsiflexion and plantar flexion.

Endoprosthetic reconstruction and rotationplasty are the most common options for joint resection in children. Some tumors are amenable only to rotationplasty, because they are large and limb salvage would not be oncologically safe. However, often, families have to make a choice based largely on how they value appearance versus durability. Although a vascularized fibula flap can salvage some joints, oncologic safety remains the priority and may prohibit this form of reconstruction.

Reconstruction With an Endoprosthesis

An endoprosthetic reconstruction is a viable option for reconstruction in patients who require resection of a joint. It can be particularly helpful for joint reconstruction in patients who prefer limb salvage and are content not to attempt high-impact activities lifelong. They should be prepared for potentially multiple revisions during their lifetime because of anticipated aseptic loosening and unanticipated complications such as infection and mechanical implant failure.

Although the use of a prosthesis can result in rapid mobilization, the hardware requires coverage with well-vascularized tissue for success. The stability of an endoprosthesis around the knee is straightforward, because the implants are constrained by a hinge. However, hemiarthroplasty using a prosthesis at the shoulder, proximal femur, and hip is difficult. It requires special consideration particularly if a wide or radical resection has removed the normally stabilizing soft tissue around the joints. Survival of an endoprosthesis has been reported to be 86%, 80%, and 69% at 3, 5, and 10 years, respectively.³⁰

Pediatric patients are more likely than adults to require revision procedures in the future in part because of device failure.² Some endoprostheses have been developed to be expandable over time to account for growth and the challenges of managing limb-length discrepancies. Most are now lengthened in a closed manner, thus limiting the number of open procedures required. However, these devices are weaker and mechanically more vulnerable than solid implants. An alternative option for endoprosthetic reconstruction in children is to use custom-sized but non-expandable implants that are solid and presumably less likely to fail mechanically. Limb length then can be managed by methods mentioned previously.

An endoprosthesis requires coverage with well-vascularized tissue to minimize infection risk, and soft tissue coverage is best undertaken at the initial surgery³¹ (Fig. 55-11). The proximal femur is deep and often has sufficient endogenous soft tissue coverage even in a wide tumor resection. The shoulder and the knee may require additional soft tissue coverage. Around the shoulder, local muscles such as the latissimus dorsi can be used, and around the knee, the medial gastrocnemius is a good option. If local muscles have been resected, free tissue transfer is required to ensure coverage of the prosthesis.

Rotationplasty

A rotationplasty is an alternative to an endoprosthetic reconstruction where the joint has been resected. This procedure is most often performed at the level of the knee for distal femur and proximal tibia tumor resections. It involves resecting a segment of the limb, including the entire knee joint, turning the remaining lower leg and ankle joint 180 degrees, and reconnecting the tibia to the proximal femur.³²⁻³⁵ Thus the ankle becomes the new knee joint³⁶ (Fig. 55-12). No joint prosthesis is required, and patients are often able to adapt remarkably well function-

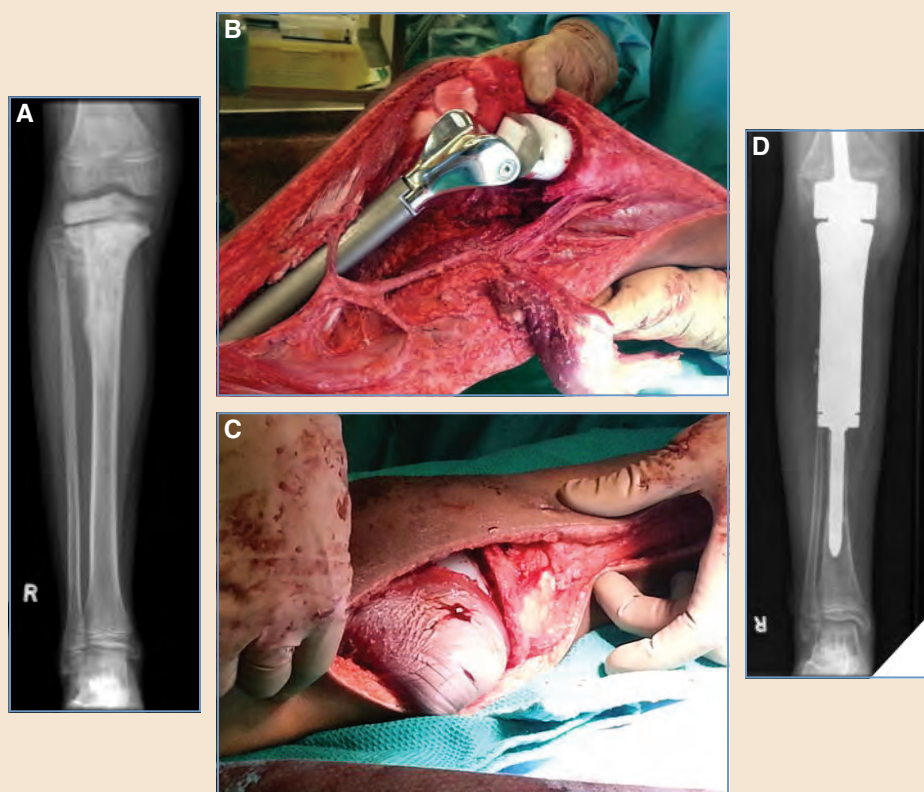


Fig. 55-11 Endoprosthesis reconstruction requires well-vascularized coverage. **A**, A preoperative radiograph showed a malignant bone tumor of the proximal tibia, necessitating joint removal. **B**, With the endoprosthesis in place, the gastrocnemius muscle was mobilized and transposed to cover the prosthetic device. **C**, The muscle flap was transferred and sutured in place. **D**, A postoperative radiograph shows the endoprosthesis well integrated into the femur proximally and the tibia distally.

ally³⁷⁻⁴⁰ (Fig. 55-13). If the vessels are safe to dissect, they are coiled, along with the sciatic nerve and its branches. To prevent arterial kinking or clotting, and if vascular supply is close to the tumor, the femoral artery and vein are resected and anastomosed to the remaining distal popliteal or posterior tibial artery and vein by the microsurgical team. Large vessel clamps are required to maintain proximal and distal control of the vessels before anastomosis. Preservation and coiling of the saphenous vein may help to prevent outflow insufficiency in the event of deep venous thrombosis. The tibial nerve is left intact to power the new knee into extension. Involvement of the tibial nerve by tumor had been regarded as a contraindication to rotationplasty. However, it is possible to resect a segment of the nerve with the tumor and coapt the clear end to successfully restore function.



Fig. 55-12 Rotationplasty for tumors of the distal femur or proximal tibia/fibula when knee joint preservation is not possible. The ankle is preserved and rotated 180 degrees. With limb shortening, it becomes the new knee joint. **A**, A preoperative MRI showed tumor invading the distal femur, precluding joint preservation. **B**, Surgical planning for a Van Nes rotationplasty. The planned incisions on the proximal thigh and leg included the biopsy site. **C**, The tumor was excised. The distal leg will be rotated 180 degrees, with the tibia connected to the femur by a plate. The sciatic nerve is intact and will be curled in continuity at the site of union. **D**, A microscope was moved into the field for repair of the femoral artery and vein to the posterior tibial artery and vein. **E**, The appearance after revascularization of the leg.

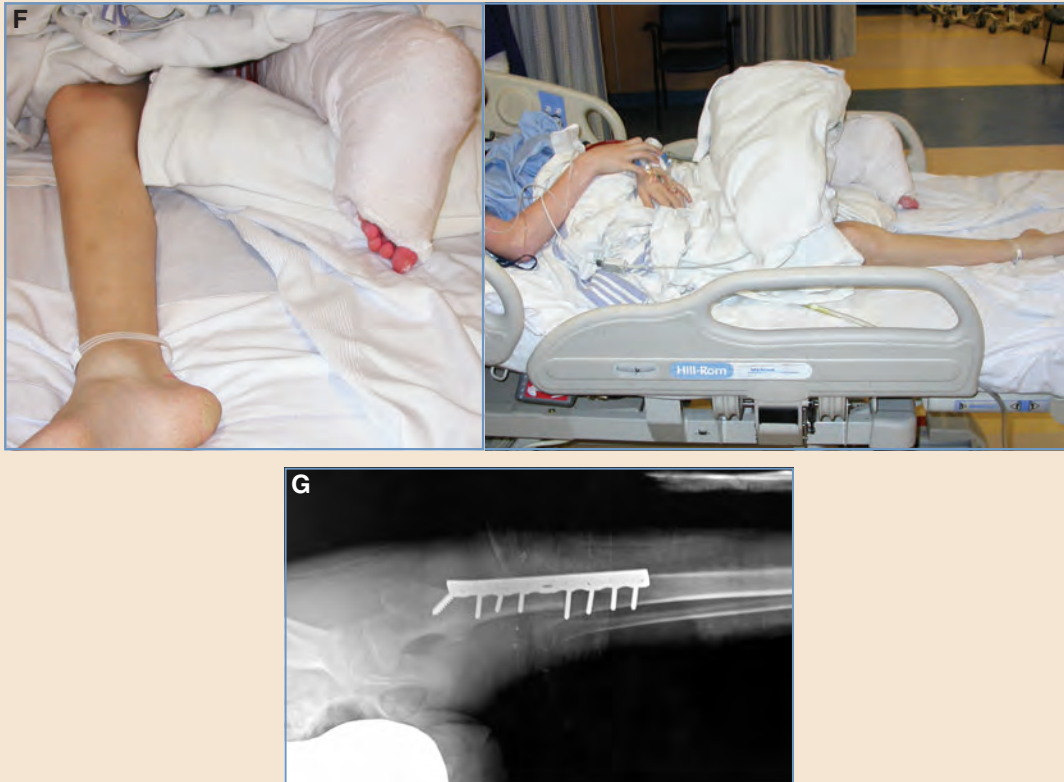


Fig. 55-12, cont'd F, The postoperative position shows rotation of leg. G, A radiograph of the femoral-tibial plating.

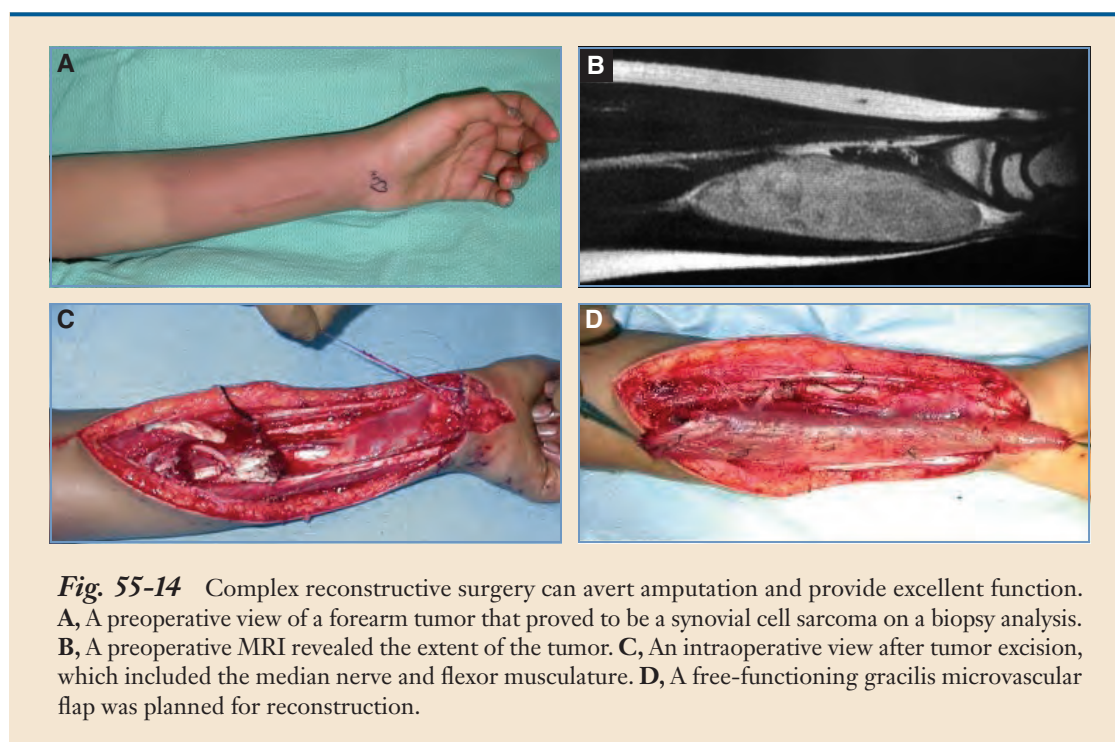


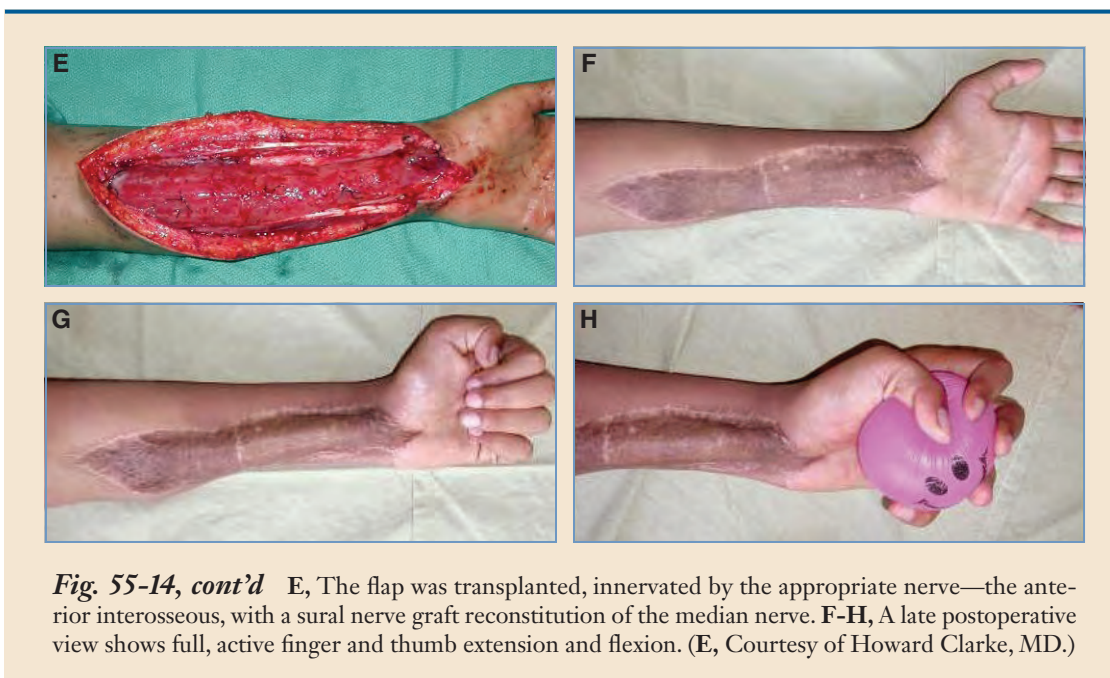
Fig. 55-13 Rotationplasty provides an excellent functional reconstruction where the knee joint must be resected. This patient underwent rotationplasty and pursued kayaking as a competitive sport. This woman was the flag bearer for Team Canada at the 2015 International Canoe Federation World Championships, where she finished seventh. She might represent Canada in the 2016 Paralympic Games also.

Controlling for leg length is an important consideration for both endoprosthesis reconstruction and rotationplasty. In a rotationplasty, the length of the reconstructed femur is typically left longer than that of the contralateral femur in growing children. This will account for future discrepant growth and gradually match the center of knee rotation by skeletal maturity. Procedures to address limb-length discrepancy include limb lengthening using distraction osteogenesis, epiphysiodesis of the long limb to limit growth, and limb shortening of the longer limb.

Amputation

In certain cases such as tumors around the ankle or foot, below-knee or more distal amputation is often the optimal procedure. For large tumors at any site, if a negative resection necessitates sacrifice of multiple critical structures, salvage may not be a viable option. Limb salvage is only reasonable if the neurovascular status of the limb is not compromised, and the function will be better than that of a prosthesis. Reconstructive techniques such as vascular and nerve grafting, nerve transfers, and functioning muscle transplantation (Fig. 55-14) continue to allow us to improve our limb-salvage abilities and avert amputation. Where amputation is necessary, creating a durable stump for future mobility and preventing neuroma formation are critical. Patient-reported functional outcome differs little between endoprosthetic reconstruction, rotationplasty, and amputation.⁴¹ However, this may not reflect patient and family preferences.





CONCLUSION

Sarcoma reconstruction in pediatric patients requires applying the principles of oncologic safety, optimizing function, and optimizing quality of life. As adjuvant therapies have improved survival, surgical techniques continue to evolve to minimize the morbidity of surgical resection. The main types of resection of tumors of the extremities include intercalary resection with vascularized bony reconstruction, joint resection with an endoprosthesis, joint resection with rotationplasty, and amputation. Free tissue transfer has significantly improved the ability to perform limb salvage surgery with good functional outcomes. Accounting for future growth of the child is an important challenge. Striving for the best possible functional outcome with the least amount of treatment burden, recognizing the potentially large number of procedures patients often require, has become the goal of reconstructive surgeons on the multidisciplinary team.

KEY POINTS

- Osteosarcoma, Ewing sarcoma, and rhabdomyosarcoma are the most common sarcomas in children.
- Tissue diagnosis requires a core biopsy that should be planned carefully along an extensile incision, with future resection in mind.
- Wide resection should be the primary surgical goal for aggressive tumors; in low-grade lesions, consideration of function may influence the excision margins.
- There are five approaches for surgical reconstruction after tumor excision:
 1. Soft tissue cover only
 2. Intercalary resection and reconstruction
 3. Joint resection and reconstruction
 4. Rotationplasty
 5. Amputation
- Vascularized fibula flaps, nerve reconstruction, and microvascular reconstruction techniques have increased the ability to salvage limbs and prevent amputation.
- The goals of treatment should be to obtain negative margins on resection and to maximize functional outcomes, while keeping the patient's and family's preferences in mind.

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56

Pediatric Lower Extremity Reconstruction

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he lower extremity presents a unique challenge to reconstructive surgeons because of its weight-bearing requirement, dependent position, subcutaneous location of the tibia, paucity of distal soft tissue, and inherent healing difficulties. As with many surgical advances, lower extremity reconstruction has significantly evolved as a result of wartime experiences, contributions of orthopedic and plastic surgeons, and advances in surgical techniques and related technologies.¹ Advances in pedicled and microsurgical flap techniques have revolutionized soft tissue reconstitution for lower extremity reconstruction.² Orthopedic advances in fixation techniques and distraction osteogenesis have also helped to expand the number of patients who are candidates for lower extremity salvage after cancer treatment and trauma.³ As the means for restoring form and function improve, indications and standards for reconstruction change. This chapter explores the issues that reconstructive surgeons face and the techniques and principles available to address these challenges.

SPECIAL CONSIDERATIONS FOR PEDIATRIC PATIENTS

Similar to reconstructive problems in adults, most reconstructive problems in children result from traumatic injuries. Congenital anomalies and defects from tumor extirpation are also encountered.^{4,7}

Pediatric patients with lower extremity defects present additional challenges, compared with adults with similar injuries.⁴ Pediatric patients and their parents may have more fear and anxiety than adult patients. Wound care and surgical and anesthetic techniques should be modified to reduce this apprehension. Examination and dressing changes may require sedation or general

anesthesia. Limited comprehension and an inability to comply with treatment recommendations further compound the difficulties of treating younger patients. However, children tend to tolerate prolonged immobilization well and generally rehabilitate more easily than adults.^{8,9} This allows a more liberal use of casts, splints, and bulky dressings, which are often used to protect surgical sites even in the absence of skeletal and ligament injury.

Pain is managed using a multimodal approach for pediatric patients with lower extremity injury. The three types of pain that often occur are (1) acute postoperative pain, (2) persistent long-term nociceptive pain, and (3) complex neuropathic pain. The rehabilitative process should include a combination of multiple pharmacologic and nonpharmacologic measures to appropriately manage pain from traumatic or surgical insult.¹⁰

A pediatric reconstructive surgeon must appreciate the dynamic nature of the structures requiring reconstruction. In patients with substantial growth potential, the impact of injury and surgeries on development must be considered. Reconstructive options that accomplish treatment goals with the least impact on development should be chosen.

EPIDEMIOLOGY

Recent statistics from the Centers for Disease Control and Prevention showed that trauma continues to be the leading cause of death among children 1 to 18 years of age, with motor vehicle accidents being a prominent factor.¹¹ An epidemiologic review of multiply injured children found that infants, toddlers, and preschoolers are most commonly injured as motor vehicle passengers, whereas children 6 to 17 years of age are more often injured as pedestrians or cyclists.¹² Traumas of the lower extremity and head are the most common types of injuries in pediatric patients with multiple injuries. Lower extremity trauma in children is also seen after all-terrain vehicle accidents,¹³ lawn mower injuries,^{8,9} and gunshot wounds.¹³

The National Cancer Institute reports that cancer incidence rates for children 0 to 14 years of age from 2008 to 2012 were 1.1 per 100,000 for soft tissues and 0.8 per 100,000 for bones and joints.¹⁴ Defects that result from excision of tumors affecting the lower extremities make up a small but challenging subset of all cases requiring lower extremity reconstruction. Combinations of soft tissue, vascular, skeletal, and neural defects, with the possibility of postoperative radiotherapy, create unique challenges in caring for these patients.

EVALUATING A TRAUMATIZED LOWER EXTREMITY

The initial management of pediatric patients with lower extremity trauma follows the Advanced Trauma Life Support guidelines established by the American College of Surgeons. Patients are assessed and treated in a systematic manner that ensures that other life-threatening injuries are detected. This prevents the treating team from being distracted by injuries that are perhaps more graphic yet less lethal than others of a more subtle nature. The initial care of a traumatized extremity may be deferred until a patient is stabilized. The evaluation consists of carefully assessing vascular, sensory, motor, and skeletal integrity and determining the extent and location of associated soft tissue defects. In many patients, sedation or even general anesthesia may be necessary for some parts of the examination. Surgeons must determine whether patients have significant bleeding or ischemia of the traumatized areas, because these issues require urgent intervention. Ischemia in the presence of a displaced fracture or a dislocation may be alleviated through reduction alone. For this reason, reduction should be performed before vascular studies and surgical exploration. Appropriate radiographic studies complement the physical examination and reveal the extent of underlying skeletal injury.

A decision to perform reconstruction or completion amputation is based on the initial viability of the tissues and the potential long-term functional outcome. In the acute setting, this decision-making can be challenging, because the extent of injury is often difficult to appreciate. For pediatric lower extremity injuries, the indications for attempted limb salvage are much more liberal than those for adults, and some authors advocate attempts at reconstruction in nearly all cases.^{4,15} Injury assessment tools have been developed to aid in decision-making. The Limb Salvage Index (Table 56-1) takes into account the degree of vascular, neural, skeletal, and soft tissue injury and warm ischemia time before presentation.¹⁶ The Mangled Extremity Severity Score is another scale of objective clinical findings that is frequently used for adults to predict the potential for salvage of a traumatized lower extremity¹⁷ (Table 56-2). In a retrospective application of this score to a pediatric trauma population, it was an accurate predictor of prognosis in 93% of injured legs.¹⁸

Early plastic surgery consultation may facilitate decision-making and earlier wound closure for patients who have lower extremity injuries with significant soft tissue defects.¹⁹ Lower extremity trauma can involve skeletal, neural, vascular, and soft tissue injuries and is best managed through a multidisciplinary approach. Restoring vascular integrity is the most important initial aspect of management after a patient is stabilized. This may involve reduction of displaced fractures or dislocations, vascular repair, or grafting. The tolerance for ischemia varies with the level of injury, the types of tissues involved, and whether the ischemia is warm or cold. Wound debridement and closure, when possible, and temporary or definitive fracture stabilization are also

Table 56-1 Limb Salvage Index

Area Assessed	Score*
Artery	0 = Contusion, palpable pulses 1 = Two-vessel leg occlusion 2 = Proximal vessel occlusion (no runoff)
Nerve	0 = Stretch injury, clean distal cut 1 = Partial transaction 2 = Complete or sciatic-level injury
Bone	0 = Closed fracture, open simple joint 1 = Multiple (>3) or open fracture 2 = Bone loss >3 cm
Skin	0 = Clean laceration 1 = Contaminated, delayed closure
Muscle	0 = Laceration, single compartment 1 = Laceration, two compartments 2 = Crush injury
Deep vein	0 = Contusion, partial laceration 1 = Complete laceration or avulsion
Warm ischemia	0 = <6 hours 1 = 6 to 9 hours 2 = 9 to 12 hours 3 = 12 to 15 hours 4 = >15 hours

*Scores greater than 6 indicate a poor prognosis for limb salvage.

Table 56-2 Mangled Extremity Severity Score

Area Assessed	Score*
Skeletal/soft tissue injury	1 = Low-energy (simple fracture, gunshot wound, stab) 2 = Medium-energy (open fracture, dislocation) 3 = High-energy (crush, high-velocity gunshot wound) 4 = Very high-energy (contaminated, open fracture)
Limb ischemia	1 = Pulse deficit, normal perfusion 2 = Pulselessness 3 = Cold, insensate (*score doubles with ischemia >6 hours)
Shock	0 = Systolic blood pressure >90 mm Hg 1 = Transient hypotension 2 = Persistent hypotension
Age	0 = <30 years 1 = 30 to 50 years 2 = >50 years

*Scores greater than 7 indicate a poor prognosis for limb salvage.

generally performed at the initial presentation. With more extensive injuries, repeated debridement is often required. Such debridement should remove clearly devitalized and contaminating material but also should preserve important functional elements when possible. Once the wound has stabilized, definitive soft tissue coverage, skeletal stabilization, and bone reconstruction can be performed.^{13,20} The preferred timing of soft tissue coverage depends on the nature of the injury. Soft tissue coverage within 7 to 15 days of injury is generally preferred.¹³ This timing has been associated with reduced infection rates in adults and generally allows technically easier surgery and lower complication rates.²¹⁻²³

HEALING BY SECONDARY INTENTION

Negative Pressure Wound Therapy

Delays in soft tissue coverage may be necessary because of factors associated with a patient's general health or because of wound-related factors. In patients with substantial contamination or extensive soft tissue injury, the need for repeated wound debridement and stabilization may prevent definitive wound coverage during the early time frame.²⁴ In these cases, lower extremity wounds can be temporized with daily wet-to-dry dressing changes or with the use of negative pressure wound therapy (NPWT). NPWT, in contrast to more traditional wound care, allows a substantial decrease in dressing changes, resulting in decreased patient pain and apprehension and the need for less sedation.²⁵ NPWT may decrease the rate of revision amputation and the need for free flap coverage; however, this may be at the expense of a longer hospitalization time.²⁶ Once the wound is debrided, generating healthy granulation tissue and adequate tissue perfusion, the NPWT can be discontinued, and either skin grafting or flap coverage can be performed, depending on the reconstructive plan.²⁶

Dermal Regeneration Templates

Dermal regeneration templates are increasingly used for coverage of lower extremity defects to minimize donor-site morbidity in pediatric patients.²⁷ It provides immediate wound coverage,



Fig. 56-1 A, This patient had a left lower extremity giant nevus extending from the middle portion of the leg to the proximal aspect of the foot. B, The affected skin was excised to unaffected deep tissues. Integra was immediately applied and left in place for 3 weeks. The outer silicone sheet was then removed, and split-thickness skin grafting was performed (not shown). C-E, The patient is shown post-operatively in an anterior view and a lateral view in dorsiflexion and plantar flexion.

even with exposed bone or tendon, when a nonautologous option is promptly required. Integra is a bilaminar dermal regeneration template comprising an outer silicone epidermis-like protection layer and a deep layer of bovine collagen and glycosaminoglycans created from shark chondroitin-6-sulfate. Once placed, Integra becomes a template for growth factors and cellular ingrowth where survival is not dependent on imbibition or inosculation.²⁷ Routine use entails placement of Integra on a clean wound bed, which becomes vascularized with cellular ingrowth over a period of 2 to 3 weeks. The silicone sheet may then be removed, and the wound is grafted or secondary intention healing is allowed to continue, potentially preventing the need for free flap coverage.²⁷ The use of dermal regeneration templates should be considered an additional tool in the reconstructive armamentarium, particularly in the difficult distal third of the leg and in clinical settings when free flap surgery may not be available (Fig. 56-1).

ANATOMIC BASIS AND SURGICAL TECHNIQUES OF LOWER EXTREMITY FLAPS

The lower extremity can be divided conceptually into regions with relatively distinct anatomic characteristics and functional properties. These areas have different reconstruction requirements and different amounts of local tissue available to lend to these efforts. The lower extremity can be considered both a donor and recipient for pedicled and microsurgically transferred flaps. Surgeons should understand the anatomy to appreciate the options and limitations of local tissue alternatives for reconstructing defects. Appreciating the physiology of the affected area helps to guide surgeons in re-creating or approximating the functional components that require repair or replacement. With the variety of techniques available, the goals of reconstruction have shifted from simply healing open wounds and eradicating infection to providing reconstructions that are both functional and aesthetic. In this section, flap options from various regions of the lower extremity are discussed. The characteristics of the flaps and their uses for reconstruction are presented.

Thigh

The thigh contains an abundance of well-vascularized soft tissue relative to the amount of bone. A number of flaps can be raised from the skin, fascia, and muscle of this region as composite flaps or isolated components and as pedicled or microsurgical flaps, depending on the reconstructive needs (Fig. 56-2).

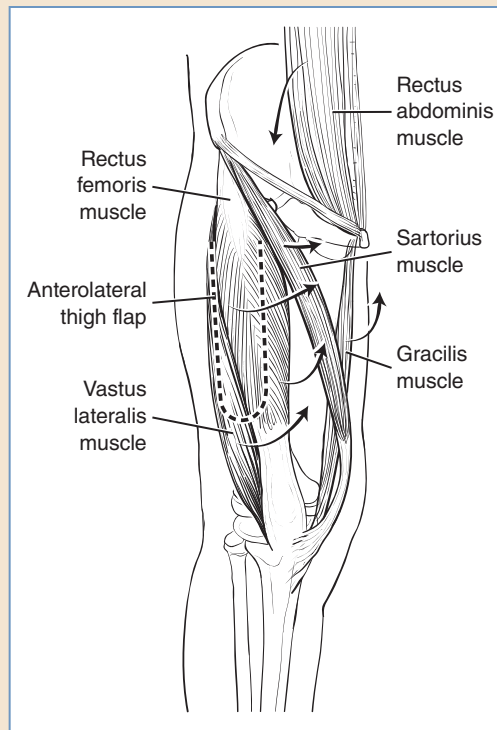


Fig. 56-2 Flap options for reconstructing the proximal thigh and groin.

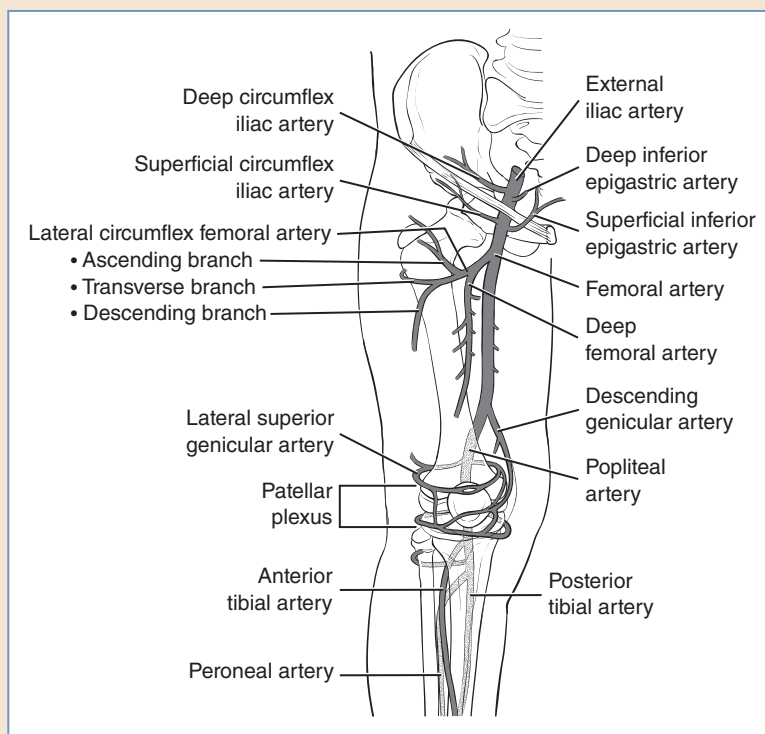


Fig. 56-3 Arterial anatomy of the lower extremity.

The skin of the thigh presents several flap choices that can be elevated as fasciocutaneous flaps. An anterolateral thigh flap is supplied by perforators from the descending branch of the lateral circumflex femoral vessels (Fig. 56-3). Most perforators are found near the central third of a line drawn from the anterior superior iliac spine to the superolateral aspect of the patella.²⁸ This line corresponds to the intramuscular septum between the vastus lateralis and the rectus femoris muscles (Fig. 56-4). Although this flap was initially described as supported by septocutaneous perforators, up to 90% of the perforators pass through the vastus lateralis muscle to supply the skin.²⁹ The skin is richly supplied through a subdermal plexus, allowing a flap to be thinned once the perforator vessels are identified and protected. A cuff of fat is left around the perforator, and the flap can be thinned to 3 to 4 mm in thickness.³⁰ Thus the flap is an option for reconstructions requiring thin flaps, even in patients with greater degrees of subcutaneous adipose tissue. An anterolateral thigh flap can be harvested as a fascial flap or raised in conjunction with the vastus lateralis muscle, the tensor fascia lata, and/or the fascia lata, depending on the reconstructive

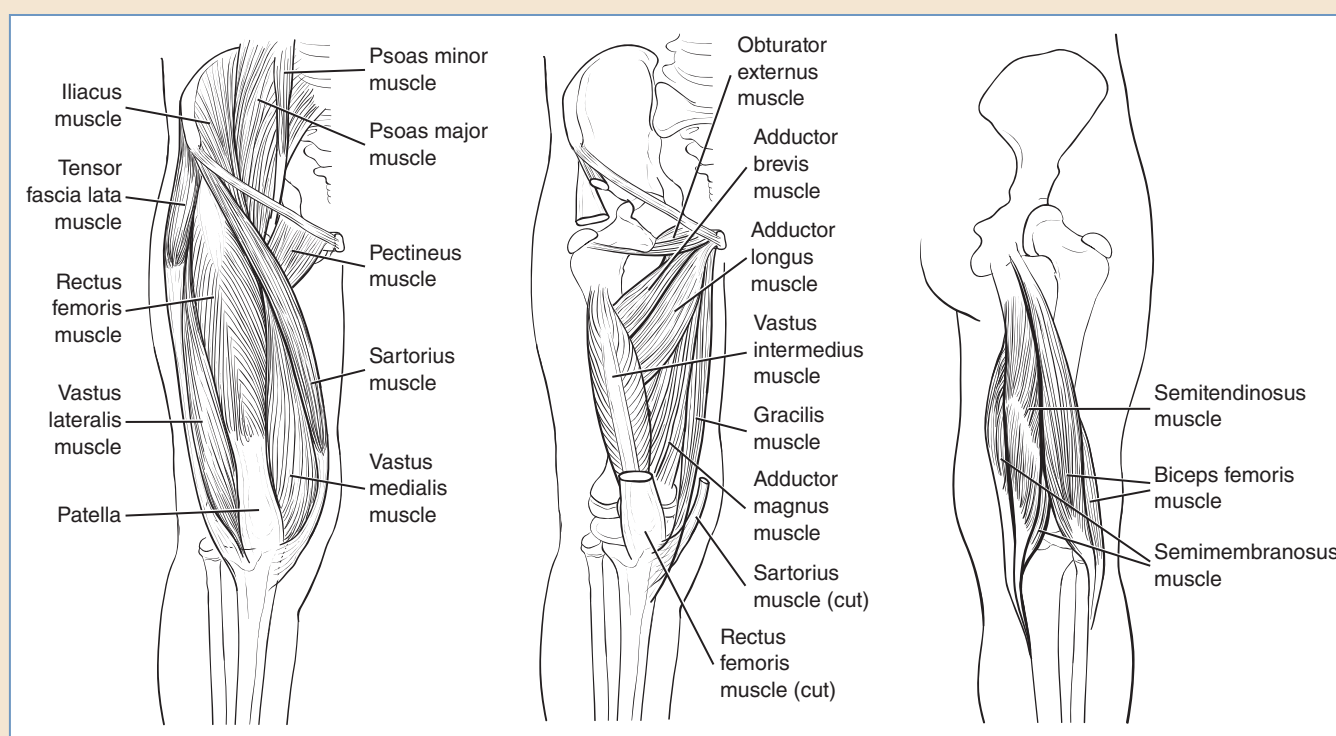


Fig. 56-4 Anatomy of the anterior thigh. This area derives vascularity primarily from deep femoral artery branches and offers a wide variety of muscle, skin, and fascia flaps.

needs³¹ (Fig. 56-5). The flap can be raised with antegrade flow through a proximal pedicle for coverage of the abdomen,³² groin, and proximal thigh or with retrograde flow through a distally based pedicle with vascular connections to the lateral superior genicular or deep femoral vessels in the distal thigh for coverage of the knee, popliteal fossa, and upper aspect of the leg³³ (see Fig. 56-3). The patellar tendon can be reconstructed simultaneously with soft tissue coverage by including fascia lata with the flap.³³ In designing these distally based flaps, surgeons must assess the adequacy of the distal vascular communications when previous surgery or trauma might have compromised them.

A lateral thigh flap is based on perforators of the deep femoral artery and may be based proximally for coverage of trochanteric and ischial wounds or distally for inferior thigh and knee defects. A medial thigh flap is based on the anterior septocutaneous branch of the superficial femoral artery in the midthigh overlying the sartorius and the rectus femoris. Applications include perineal wounds, genitourinary reconstruction, and postburn groin contractures.³⁴

The muscles of the thigh comprise a substantial amount of soft tissue divided into three fascial compartments (Box 56-1; see Fig. 56-4).

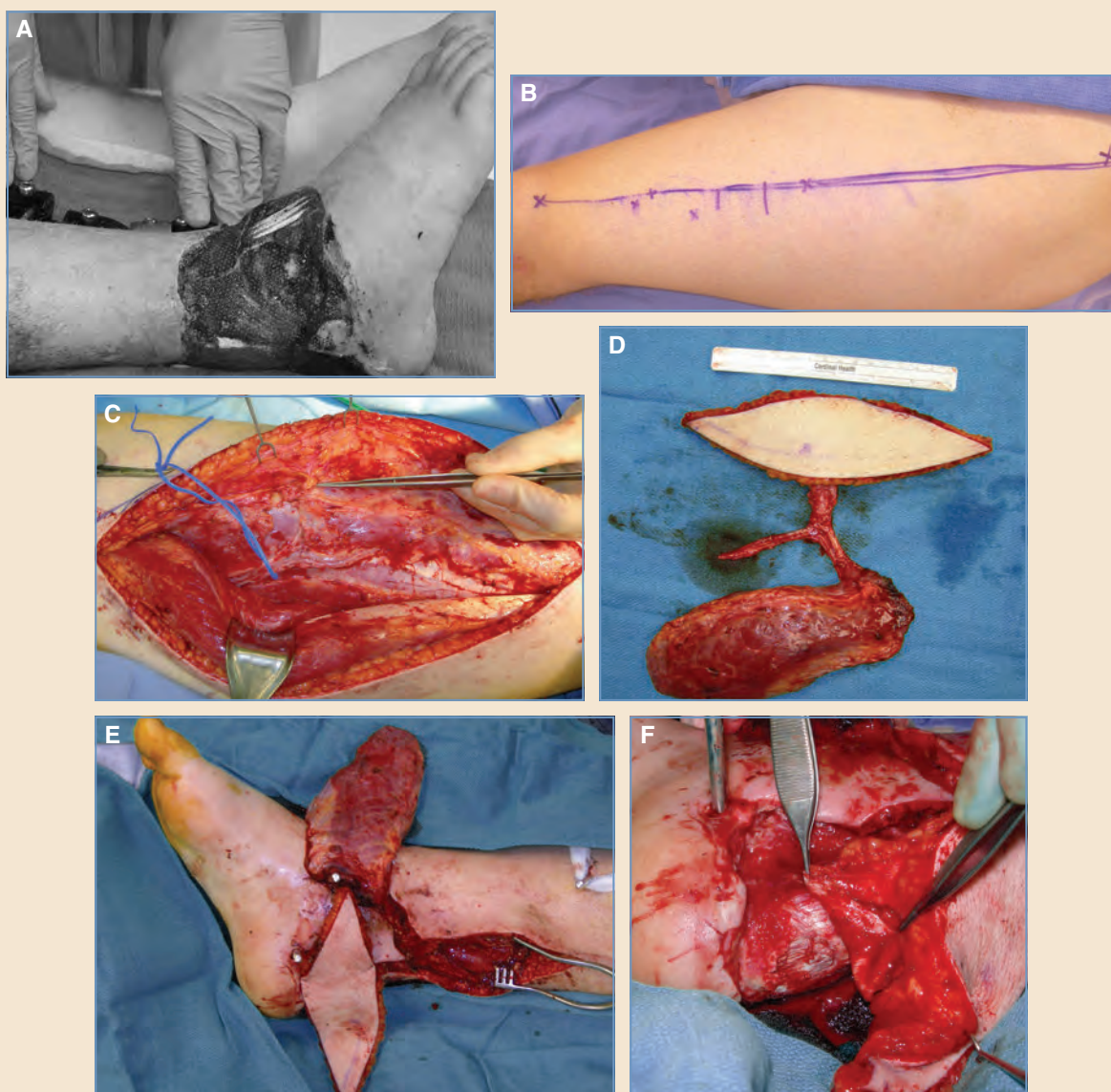


Fig. 56-5 **A**, This patient had an open dislocation of the right ankle with loss of soft tissue coverage. The wound was nearly circumferential. The ankle joint was stabilized with an external fixator. **B**, The intermuscular septum between the vastus lateralis and rectus femoris muscles of the left thigh was marked. Cutaneous vessels were identified with a handheld Doppler probe and their positions marked. **C**, The descending branch of the lateral circumflex femoral artery was identified between the vastus lateralis and rectus femoris muscles. The vessel penetrated the vastus lateralis muscle as it coursed distally in the thigh. A branch that coursed to the skin was identified within the intermuscular septum. **D**, An anterolateral thigh flap and a partial vastus lateralis flap were raised from the left thigh with a common pedicle (the descending branch of the lateral circumflex femoral artery and vein). Generous pedicle length is a useful feature of these flaps. **E**, Anastomosis to the posterior tibial vessels was performed, and a flap inset was planned. **F**, A segment of fascia lata included on the deep surface of the skin paddle was used as a vascularized fascia flap for reinforcement of the compromised Achilles tendon.

Continued



Fig. 56-5, cont'd **G**, A lateral view of the leg after flap inset. **H**, A medial view of the leg after flap inset. **I**, The thigh donor site after primary closure. **J-N**, The healed ankle wound. Muscle debulking and skin grafting were performed in the interim.

Box 56-1 Fascial Compartments and Muscles of the Thigh**Anterior Compartment (innervated by the femoral nerve)**

- Rectus femoris
- Vastus medialis
- Vastus lateralis
- Vastus intermedius
- Tensor fascia lata
- Sartorius
- Iliacus
- Psoas
- Pectineus

Medial Compartment (innervated by the obturator nerve)

- Gracilis
- Adductors (longus, brevis, and magnus)
- Obturator externus

Posterior Compartment (innervated by the sciatic nerve)

- Biceps femoris
- Semitendinosus
- Semimembranosus

The vascular supply to the muscles of the thigh is primarily derived from branches of the deep femoral artery (see Fig. 56-3). Most flaps in this region are based on branches of the lateral and medial circumflex femoral artery. Lateral muscle groups are supplied by the transverse, ascending, and descending branches of the lateral circumflex femoral artery, whereas the adductors and the gracilis are supplied by the medial circumflex vessels.³⁵ Perforating branches of the lateral circumflex femoral artery and the deep femoral artery supply the skin territories of the anterolateral and lateral thigh. The superficial femoral artery provides a segmental blood supply to the sartorius muscle and perforator vessels that supply the anteromedial thigh skin. The obturator and inferior gluteal vessels provide vascular supply to the skin of the medial and posterior thigh.^{8,36} As the superficial femoral artery approaches the knee through the adductor hiatus, it becomes the popliteal artery, giving off the sural, genicular, and saphenous branches.

With abundant surrounding soft tissue, most thigh defects can be closed by local tissue advancement or skin grafting. Large defects and wounds with exposed femoral vessels or femoral fractures, with or without exposed hardware, may occasionally require regional or even free flap coverage.³⁶ Pedicled rectus abdominis muscle and vertical rectus abdominis myocutaneous flaps are useful for large anterior thigh defects and coverage of exposed femoral vessels.³⁷ Regional flaps available for closure of proximal thigh, hip, and groin wounds include vastus lateralis, rectus femoris, and tensor fascia lata muscle flaps.

The vastus lateralis is a broad, thick muscle located deep to the tensor fascia lata in the lateral thigh. It can be harvested as a muscle flap or as a myocutaneous flap through inclusion of anterolateral thigh skin supplied by perforators through the muscle. Its harvest has little effect on ambulation, because the remaining muscles of the quadriceps group continue to function for knee extension.³⁸ The flap is supplied by the descending branch of the lateral circumflex femoral vessels. It is particularly useful for coverage of trochanteric and hip defects,³⁹ but it is also effective as a distally based flap³³ (Fig. 56-6) or a microvascular flap.⁴⁰



Fig. 56-6 A, This patient was involved in a snowmobile accident and presented with a lateral knee wound. The lesion measured 15 by 16.5 cm. B, Knee flexion revealed an open knee joint and an exposed patellar tendon. C, A distally based vastus lateralis muscle flap was harvested. D, Careful assessment of venous drainage is important with distally based flaps. A “supercharge” of venous outflow through anastomosis of a vena comitans of the descending branch of the lateral circumflex femoral artery with the large saphenous vein prevented venous congestion. E, The inset muscle provided complete joint coverage. F, The muscle flap was covered with a skin graft.

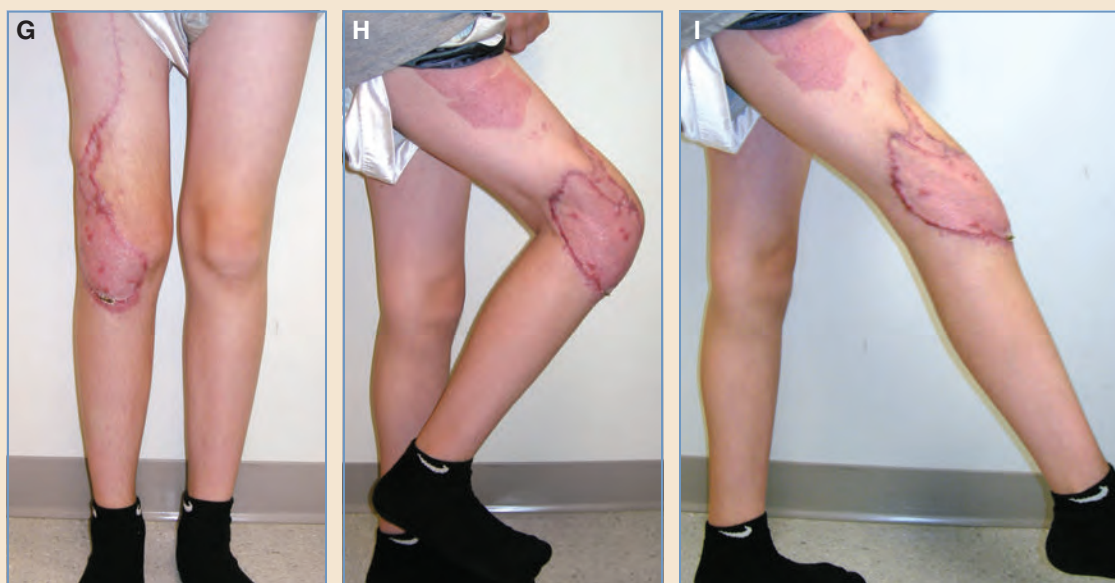


Fig. 56-6, cont'd G-I, The healed wound.

The rectus femoris muscle, located centrally in the anterior thigh, receives its vascular supply primarily from the descending branch of the lateral circumflex femoral vessels. Branches of the superficial femoral vessels and the ascending branch of the lateral circumflex femoral vessels may provide additional minor contributions.⁴¹ This muscle is not as bulky as the vastus lateralis and can be elevated as a myocutaneous flap with a sizeable skin paddle. Despite concerns that its harvest may adversely affect knee extension, reports show that other muscles of the quadriceps group compensate well in its absence.⁴²

The tensor fascia lata muscle, located in the lateral thigh, is a thin, flat muscle with numerous regional and microvascular applications. Based on the ascending branch of the lateral circumflex femoral vessels, this muscle can be elevated as a muscle, myocutaneous, myotendinous, or osteomyocutaneous flap.⁴³ It can also be raised in combination with the rectus femoris for large defects, as in a “muttonchop” flap.⁴⁴ The cutaneous portion of this flap receives T12- and L1-level innervation from the lateral femoral cutaneous nerve and branches of T12. With preservation of these nerves, sensation can be transferred with the flap. This can be useful for covering posterior pelvic defects where a lack of sensation can lead to recurrent pressure ulceration. In the thigh, this flap is used most frequently for coverage of trochanteric and ischial pressure sores but can also be applied to groin defects.^{45,46}

The sartorius muscle is supplied by segmental perforators of the superficial femoral vessels. Vascular communications between segmental territories allow the muscle to be elevated as a pedicled flap.⁴⁷ It is a long, thin muscle used most often in the thigh as a turnover flap for covering complicated wounds after vascular bypass.³⁷ It can also be advanced superiorly over groin defects or inferiorly over lateral knee defects.⁴⁸

The biceps femoris muscle is the lateral component of the hamstring muscle group of the posterior thigh. It is supplied by branches of the deep femoral vessels. It is used primarily for closure of ischial pressure ulcers sores,⁴⁹ but it can also be employed for defects of the anterior thigh and functional reconstruction of the quadriceps.⁵⁰

The gracilis muscle is a member of the adductor group of muscles, located along the medial aspect of the thigh between the adductor longus and semimembranosus muscles. It is supplied by a reliable branch of the medial circumflex femoral vessels that runs between the adductor longus and magnus muscles and by minor branches of the superficial femoral artery. It is effective as a pedicled or free muscle flap. Because of its reliability, it is used frequently for distal lower extremity defects in pediatric patients.⁵¹ The overlying skin can be included in transverse or longitudinal orientation relative to the muscle.⁵² The muscle is supplied by a well-characterized branch of the obturator nerve that accompanies the vascular pedicle as it approaches the muscle. The muscle is useful as an innervated flap for functional reconstruction of facial nerve paralysis and for upper and lower extremity defects.⁵³

Distal Thigh and Knee

Soft tissue availability significantly decreases distally in the thigh, resulting in greater challenges in designing local coverage options for supracondylar and knee defects. Thigh-based coverage options include regional muscle flaps, such as the sartorius and vastus medialis muscles, and fasciocutaneous flaps, including distally based anterolateral and anteromedial thigh flaps. Generally, the best regional tissue options are the gastrocnemius and soleus muscles. Fasciocutaneous flaps from the leg based on the saphenous artery and superficial branches of the superficial femoral artery have also been described.³⁵ For larger defects and in patients in whom these regional flap options are unavailable, microvascular tissue transfer is usually required (Fig. 56-7).

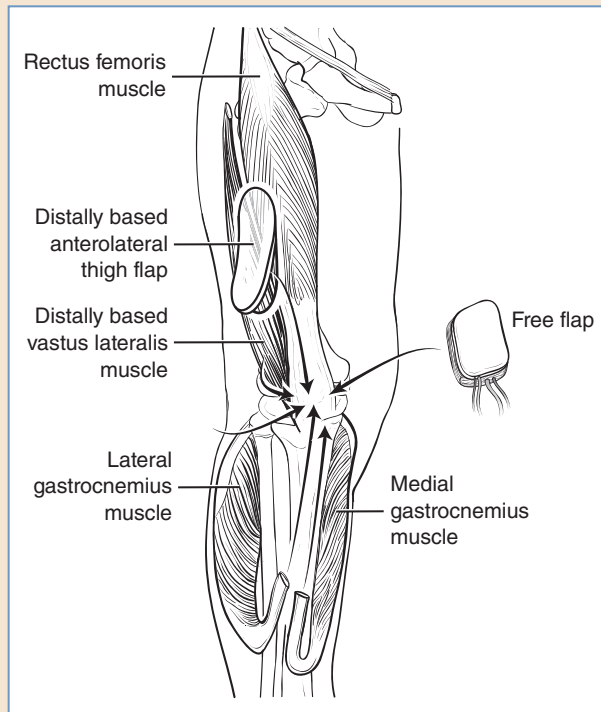


Fig. 56-7 Flap options for reconstruction of the distal thigh and knee.

Leg

The leg comprises four fascial compartments that contain the musculature and neurovascular structures (Fig. 56-8). The anterior compartment contains the tibialis anterior, extensor digitorum longus, extensor hallucis longus, and peroneus tertius muscles. It also contains the anterior tibial vessels and the deep peroneal nerve, which supply these muscles. The lateral compartment contains the peroneus longus and brevis muscles, which are innervated by the superficial peroneal nerve. The posterior compartment is subdivided into superficial and deep parts. The superficial portion contains the gastrocnemius, soleus, and plantaris longus muscles. The deep portion contains the flexor digitorum longus, the flexor hallucis longus, the popliteus and tibialis posterior muscles, the peroneal and posterior tibial vessels, and the tibial nerve. All muscles of the posterior compartment are innervated by the tibial nerve.

The popliteal artery gives off a number of branches, including the sural arteries that supply the gastrocnemius muscle and the genicular arteries (see Fig. 56-3). The popliteal artery further divides into the anterior tibial artery and a common trunk, which subsequently divides into the peroneal and posterior tibial arteries. The anterior tibial artery courses through the interosseous membrane to supply the muscles of the anterior compartment and the anterolateral skin of the leg. At the foot it becomes the dorsalis pedis artery, which contributes to the deep plantar arch. The posterior tibial artery supplies the skin of the posteromedial aspect of the leg and the muscles of the posterior compartment. In the foot, it divides to become the medial and lateral

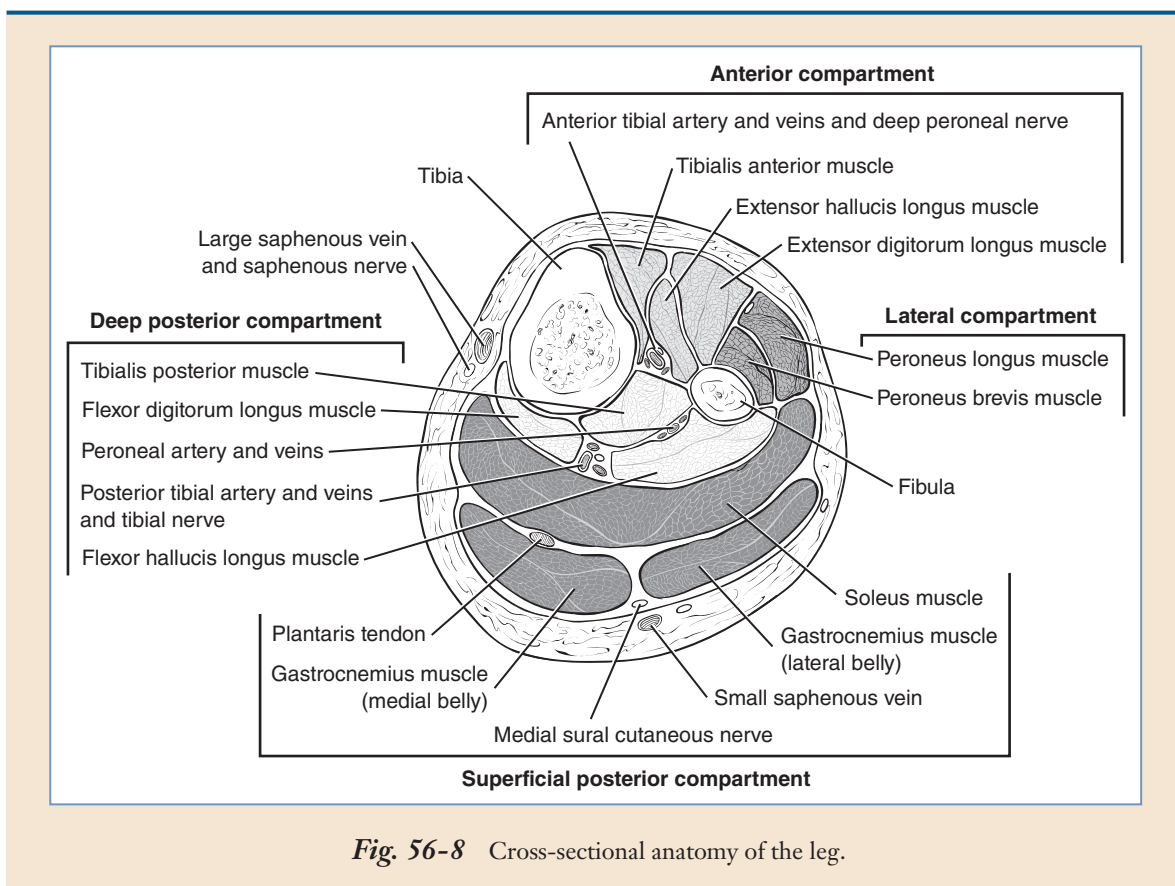


Fig. 56-8 Cross-sectional anatomy of the leg.

plantar arteries. The peroneal artery supplies the muscles of the lateral compartment, the fibula, and the posterolateral skin of the proximal and middle parts of the leg.⁵⁴

Defects of the proximal and middle parts of the leg are usually managed with fasciocutaneous flaps or muscle flaps from the gastrocnemius or soleus muscles. Fasciocutaneous flaps based on the peroneal, anterior and posterior tibial, and sural arteries have been described.^{55,56} In general, muscle flaps in the leg are covered with skin grafts rather than elevated as myocutaneous flaps to reduce donor-site morbidity and to aid in incorporating the flap into the defect.

Supplied by the medial and lateral sural arteries, the gastrocnemius muscle is the workhorse flap for defects of the supracondylar area, the knee, and upper third of the leg. It is located superficially in the posterior calf and is divided into medial and lateral heads, both of which can be used as independent muscle flaps. The medial head is generally preferred for flap purposes, because it is longer and its use prevents dissection around the peroneal nerve, which overlies the origin of the lateral head in the subcutaneous tissue. The origin of either muscle can be divided and the vascular pedicle dissected into the popliteal fossa to increase the arc of rotation or advancement.⁵⁷ For larger defects both heads can be used, or one head can be used in combination with a soleus muscle flap. At least one of the three motor units must be preserved to maintain plantar flexion at the ankle. If the defect is small, a portion of the medial head may be returned to the donor site and reattached to the lateral head and the Achilles tendon for improved function.⁵⁸ The gastrocnemius may be elevated as a myocutaneous flap with a skin-fascial extension⁵⁹ or raised as an island flap that may reach the middle to distal femur.⁶⁰

The soleus muscle lies deep to the gastrocnemius muscle in the superficial posterior compartment. It originates from both the fibula and tibia and inserts into the deep surface of the Achilles tendon. The muscle can be split (hemisoleus flap) or transposed whole for coverage of the middle third of the leg.²

Although free tissue transfer has become routine for covering distal lower leg and foot defects in adults and children, local coverage options have been described and are used in many cases. Fasciocutaneous coverage includes distally based variants of the peroneal, anterior and posterior tibial, and sural artery flaps.⁶¹ These flaps can leave significant donor defects and may require a microvascular anastomosis to ensure adequate venous drainage. Axial fascial and adipofascial flaps limit donor defects but provide only thin coverage.⁶² Turnover adipofascial flaps are useful for coverage of proximal and distal defects. These flaps are not limited by the location of perforators and can be oriented horizontally or vertically, with a mobilized length/width ratio of up to 3:1⁶³ (Fig. 56-9).

The extrinsic extensors and flexors of the toes and ankle have been described for muscle coverage of the distal tibia and ankle region, but these are all small muscles with limited local usefulness.⁶⁴ The reversed hemisoleus muscle flap, based on a perforator from the posterior tibial artery, has also been described for distal leg coverage, but its utility is limited by problems with reliability.⁶⁵

Foot

The dorsum of the foot is covered by thin fascia that is continuous with the transverse and cruciate crural ligaments proximally and the plantar fascia medially and laterally (Fig. 56-10). Distally this fascia forms the sheath of the extrinsic extensor tendons. Deep to the long extensor tendons, the extensor digitorum brevis arises from the superior and lateral aspects of the calcaneus. This thin, broad muscle is supplied by the lateral tarsal branch of the dorsalis pedis artery and innervated by the deep peroneal nerve.

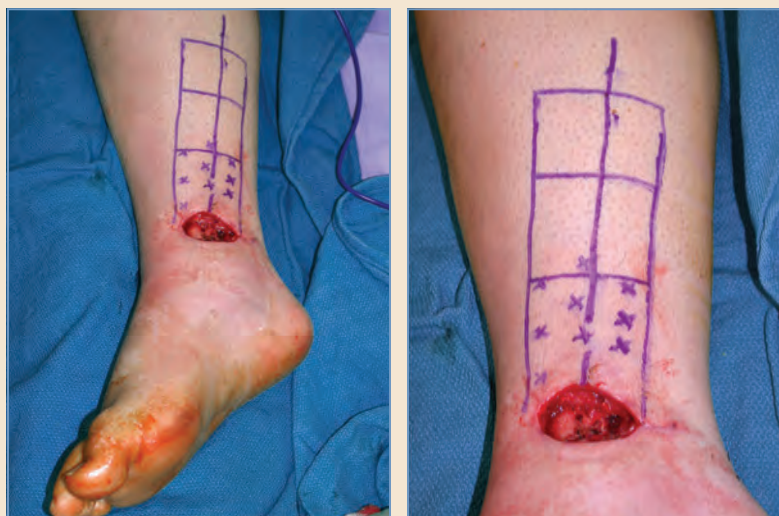


Fig. 56-9 Design of an adipofascial turnover flap. A handheld Doppler probe is used to identify perforating vessels, and the flap base is centered over this area. The total flap length is three times the length of the defect to be covered. The distal third covers the defect, the middle third is folded on itself, and the proximal third is the flap base. The fascia and fat of the flap are dissected from the overlying skin and underlying muscle. The skin is closed primarily, and the flap is covered with a skin graft. (Courtesy of Michael L. Bentz, MD.)

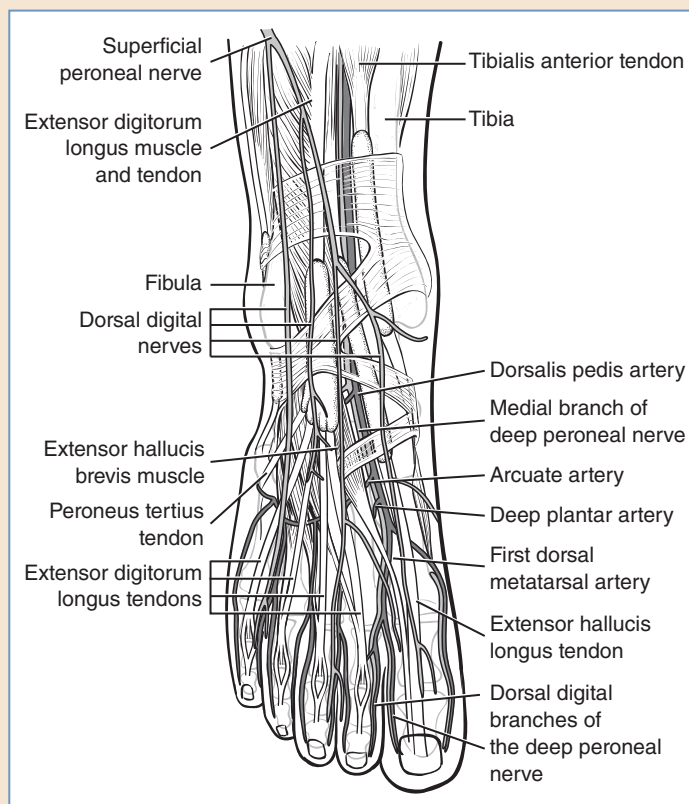


Fig. 56-10 Anatomy of the dorsum of the foot.

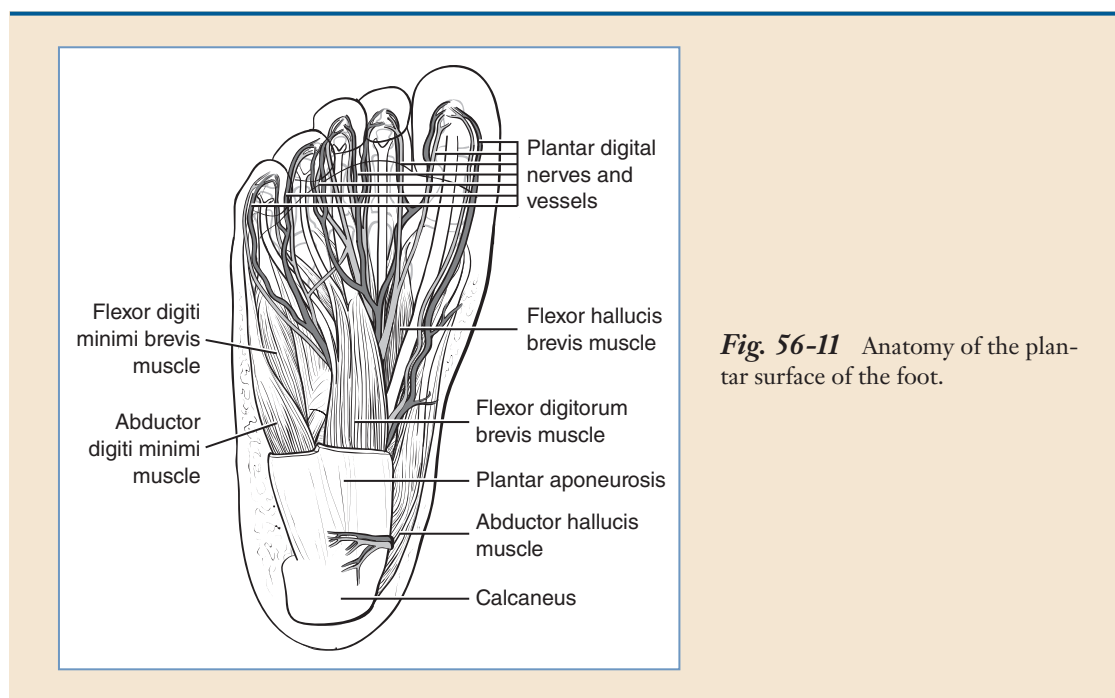


Fig. 56-11 Anatomy of the plantar surface of the foot.

The plantar surface of the foot has unique properties related to its function (Fig. 56-11). It is covered by glabrous skin, which is extraordinarily thick over the calcaneus and anchored to the underlying plantar fascia by thick septi, interspersed by loculations of semisolid fat. The plantar fascia extends from the calcaneus distally, and inserts into the skin and the sheaths of the flexor tendons. Superficial muscles of the plantar surface include the abductor hallucis and the flexor digitorum brevis, innervated by the medial plantar nerve. The abductor digiti minimi is located superficially on the lateral aspect of the foot and is innervated by the lateral plantar nerve. Muscles deep to the plantar vessels and nerves include the quadratus plantae, lumbricals, abductor digiti quinti, adductor hallucis, flexor hallucis brevis, and flexor digiti minimi brevis.

The vascular supply of the foot is derived from the continuation of the vessels of the leg (see Fig. 56-3). The anterior tibial artery becomes the dorsalis pedis artery, which gives off the medial and lateral tarsal arteries and the first dorsal metatarsal artery and continues as the deep plantar artery. The posterior tibial artery gives off the medial calcaneal artery before dividing into the medial and lateral plantar arteries, deep to the origin of the adductor hallucis. The lateral plantar artery unites with the terminal dorsalis pedis artery to complete the deep plantar arch. The peroneal artery terminates in the foot as the lateral calcaneal artery.

Large defects of the distal ankle and foot usually require free tissue transfer for coverage. A number of local flap options exist for treating small- to medium-sized defects in many areas. Distally based fascial or fasciocutaneous flaps from the leg or fasciocutaneous flaps supplied by the dorsalis pedis, medial and lateral plantar, and lateral calcaneal arteries may be used to cover certain defects.^{66,67} Muscle flaps useful for coverage of selected distal defects include the extensor digitorum brevis, abductor hallucis, abductor digiti minimi, flexor digitorum brevis, and flexor digiti minimi muscles⁶⁶ (Figs. 56-12 and 56-13).

The dorsalis pedis flap is a thin fasciocutaneous flap that can be raised as a proximally or distally based local flap or as a microvascular flap.^{68,69} Its pedicle is the dorsalis pedis artery and the venae comitantes when based proximally⁶⁷ and the first dorsal metatarsal artery and the venae comitantes when based distally.⁷⁰ Saphenous vein branches can provide venous drainage when

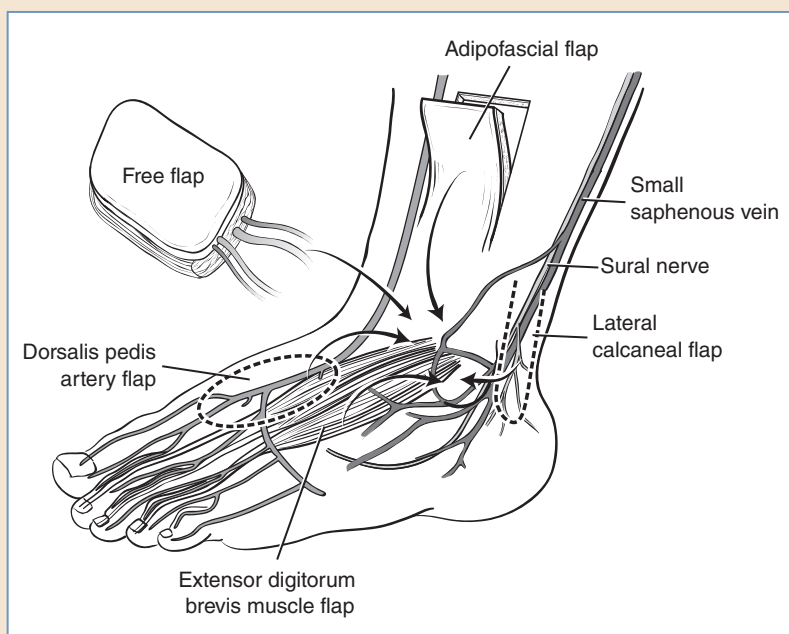


Fig. 56-12 Flap options for ankle reconstruction.

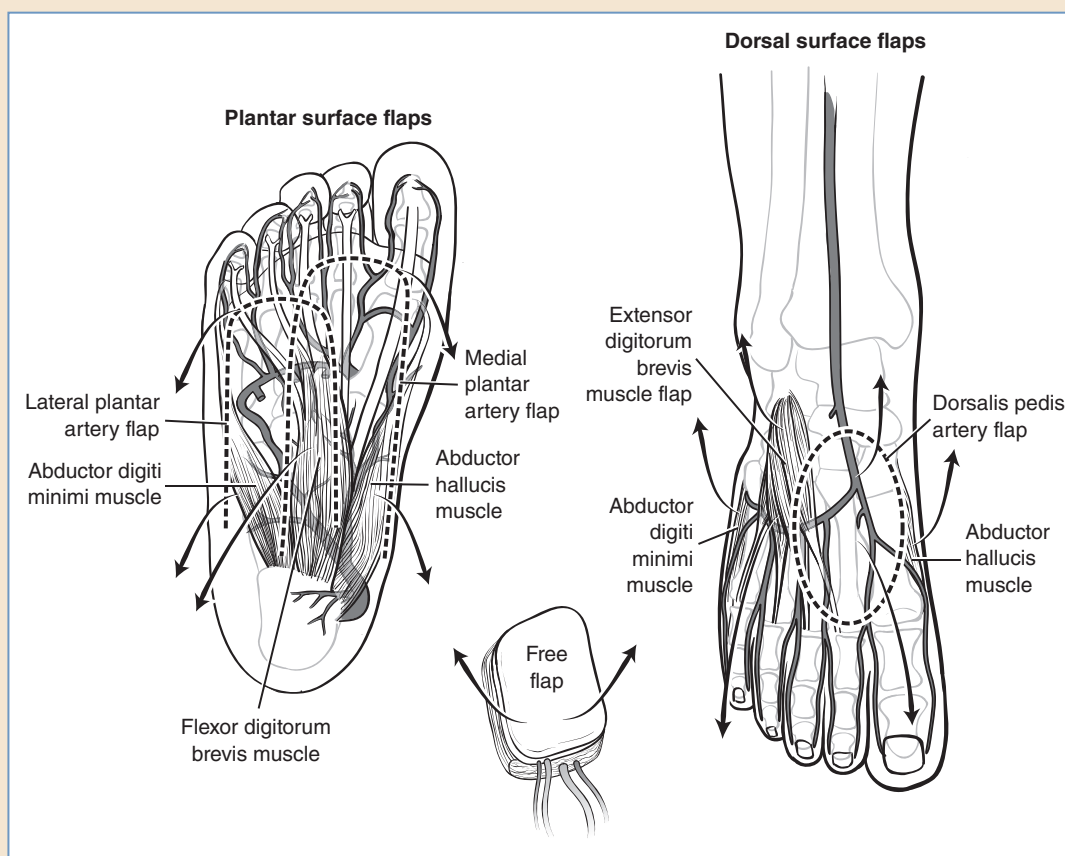
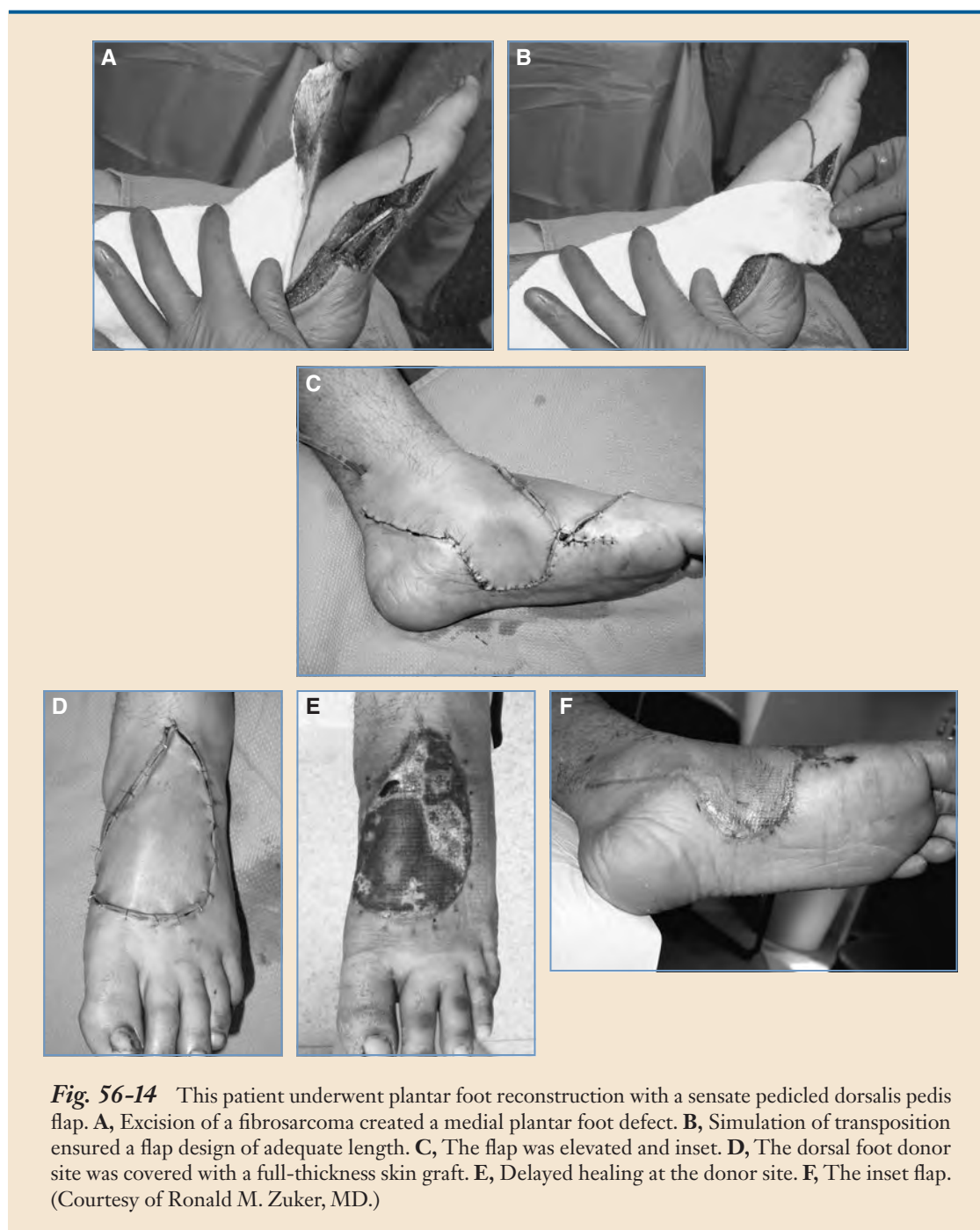


Fig. 56-13 Flap options for dorsal and plantar foot reconstruction.

the flap is used during microsurgery. Flap modifications have been described, including the inclusion of the vascularized tendon and extensor digitorum brevis muscle,⁷¹ elevation of the flap as a myofascial or adipofascial flap,⁷² inclusion of the second toe or metatarsophalangeal joint,⁷³ and neurotization of the flap through the deep or superficial peroneal nerves.^{68,69} The flap can be employed to provide sensate coverage of the plantar surface of the foot as either a pedicled or microvascularly transplanted flap⁷⁴ (Fig. 56-14). Problems of donor-site morbidity have been



reported, including delayed healing and wound breakdown, pain, scarring, contracture, and infection.⁷⁵ The incidence and severity of these problems can be reduced with meticulous dissection of the donor site, preservation of the paratenon on tendons,⁷⁶ and the use of the extensor digitorum brevis muscle for tendon coverage.⁷⁷

The extensor digitorum brevis can be harvested with antegrade or retrograde flow through the dorsalis pedis artery to the lateral tarsal branch that supplies the muscle. Proximally based flaps are well described for covering wounds of the proximal part of the foot and the ankle.⁷⁸ The extensor digitorum brevis can also be included in the fasciocutaneous territory of the dorsalis pedis artery for a myocutaneous flap.⁷⁹ The distally based flap is useful for coverage of the middle to distal foot.⁸⁰ The extensor digitorum brevis is harvested through a dorsal incision lateral to the dorsalis pedis vessels. The lateral tarsal arterial branch is identified between the extensor digitorum longus and extensor hallucis longus tendons and is traced into the muscle. The dorsalis pedis artery may be ligated distally or proximally as needed to improve the arc of rotation. Before ligation of the artery, the integrity of the posterior tibial circulation must be confirmed to prevent vascular compromise of the foot. The superficial surface of the muscle is dissected, preserving the long extensor tendons and the superficial peroneal nerve. The short extensor tendons are divided, and the deep surface is dissected off of the tarsal bones. The sinus tarsi branch from the lateral tarsal artery must be identified and divided to prevent injury to the pedicle.

The lateral calcaneal artery flap is based on the terminal branches of the peroneal artery located between the Achilles tendon and peroneal tendons (Fig. 56-15). The flap is innervated by the sural nerve. This flap has been described with various modifications for Achilles tendon coverage and reconstruction of calcaneal and plantar heel defects, including as a transposition flap, island flap, and V-Y advancement flap.⁸¹ This flap is useful for transferring durable, sensate tissue without sacrificing the plantar arteries or nerves.

Heel defects are difficult to reconstruct because of the specialized tissue requirements of sensation and durability. Skin grafts, although frequently suboptimal, can be used in any portion of the plantar foot as long as adequate subcutaneous tissue is present.⁸² Sensate flaps are ideal for heel coverage and can be provided by the calcaneal artery flap, including the medial calcaneal nerve, or the medial plantar artery fasciocutaneous flap, which includes the medial plantar nerve. Hidalgo and Shaw^{83,84} described the rotation of a medially based flap of heel tissue that preserves both the medial and lateral plantar nerves. Intrinsic muscle flaps, including the flexor



Fig. 56-15 Design of a lateral calcaneal artery flap. The position of the artery is confirmed with a handheld Doppler probe. The flap is elevated with inclusion of the artery and can be used for coverage of the lateral malleolus or Achilles tendon. The donor site is covered with a skin graft. (Courtesy of Michael L. Bentz, MD.)

digitorum brevis and the abductor hallucis brevis, are useful for small- to moderate-sized and deeper defects.⁸⁵ Their coverage is limited because of their relatively small size and limited potential for advancement. Large defects and those with exposed hardware or bone often require free tissue transfer.⁸⁶ Muscle flaps are usually indicated for larger defects and when osteomyelitis is present. Innervated fasciocutaneous flaps are best for acute defects when sensation could be valuable.^{86,87} Muscle flaps are generally preferred over fasciocutaneous flaps for plantar coverage, because they provide greater stability and create less shear between the flap surface and the deep tissues with walking; however, this is debatable. Both types of flap show good function over time, and selection should be based on the patient's characteristics and needs.⁸⁷ Noninnervated flaps show some degree of sensory recovery, although it does not reach the level of two-point discrimination or light touch.⁸⁸

Dorsal foot defects can be covered with skin grafts if the paratenon is preserved or if exposed tendons are excised (Fig. 56-16). Larger wounds, particularly with exposed bone, usually require free flap coverage. Ideally the tissues used are thin, pliable, and durable.⁸⁹ Coverage of distal defects with local flaps such as reverse-flow dorsalis pedis and extensor digitorum brevis flaps, based on the lateral tarsal or first dorsal metatarsal arteries, has also been described.^{70,79}



Fig. 56-16 This patient had a distal dorsal foot wound resulting from a lawn mower accident. This case demonstrates complex wound coverage using a sequential approach to wound management. **A**, Initial presentation with exposed bone and tendon. **B**, Appearance after local flap coverage of tendon and bone. **C**, Appearance after NPWT therapy for 3 days. **D**, Appearance after split-thickness skin grafting. **E**, The healed wound.

Distal plantar defects are best covered with local sensate flaps, such as fillet flaps from central digits or small V-Y perforator flaps.⁹⁰ Distally based muscle and fasciocutaneous flaps have been described, including the abductor digiti minimi, flexor hallucis brevis, and medial plantar flaps. Non-weight-bearing portions of the plantar foot can be favorably managed with skin grafts.

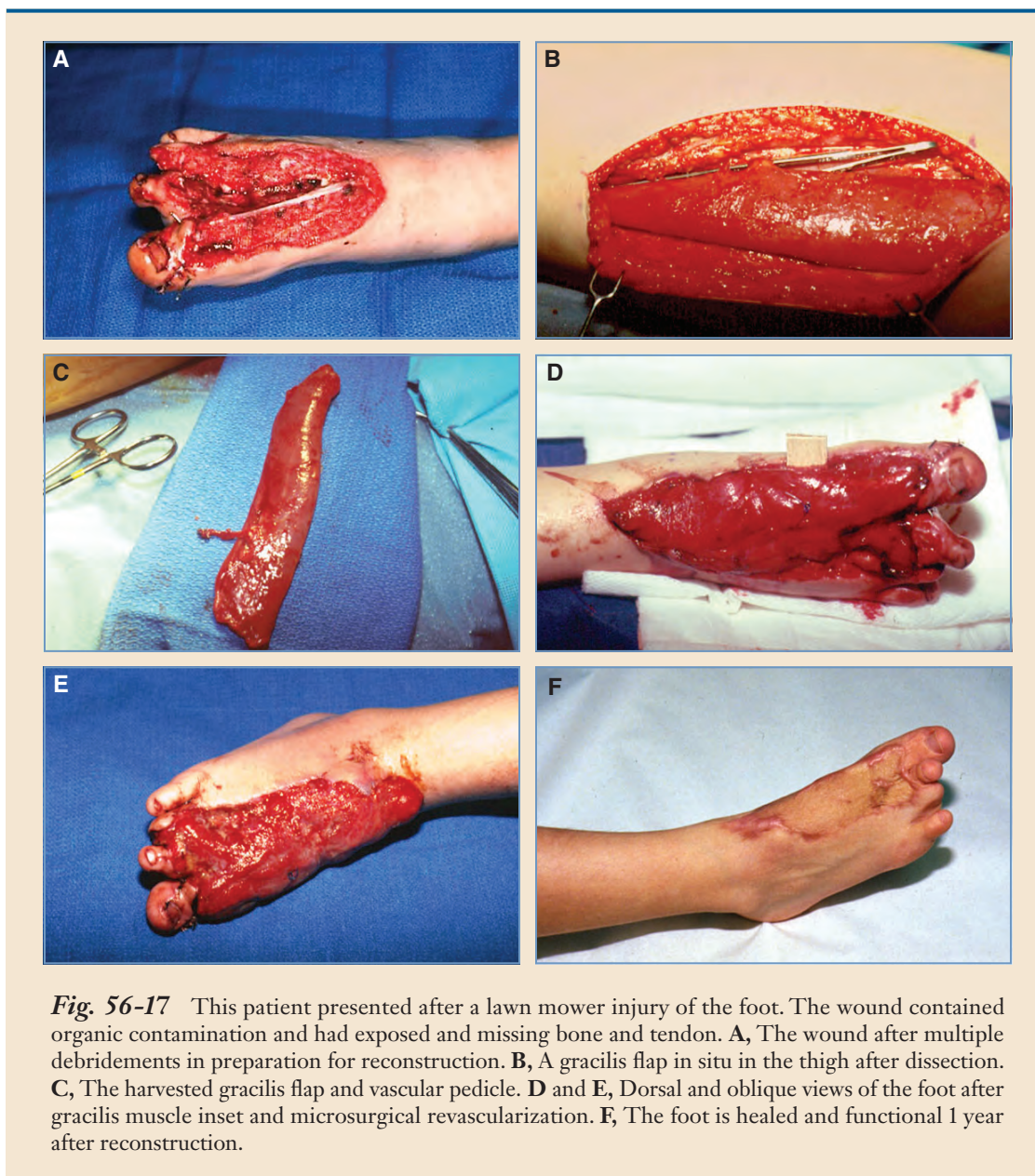
MICROSURGICAL TRANSPLANTATION

Microvascular free flaps have become the benchmark for covering difficult defects of the lower extremity.^{6,91,92} Indications for microvascular reconstruction include high-energy injuries, many middle and most distal tibial defects, radiation-related wounds, tumor reconstruction, and osteomyelitis; it is also useful for part of the treatment for skeletal nonunion. Over the past 30 years, microsurgery has achieved a level of sophistication that has refocused the reconstructive goals from tissue survival and coverage to improvement in appearance, limitation of donor-site morbidity, and preservation of function.

Hariri and Ohmori⁹³ originally reported on pediatric free tissue transfer in 1975. They harvested free groin flaps for coverage of ankle defects in two 4-year-old children. Since then, numerous applications of microvascular flaps in children have been described, ranging from toe-to-thumb transfer to esophageal reconstruction with jejunal segments.⁹⁴ Congenital problems, such as congenital hand deformities,⁵¹ account for a significant portion of free tissue transfers performed in children, but lower extremity trauma requiring free flap closure in pediatric patients has been reported.^{91,95,96} In 2008 Upton and Guo⁹¹ reported on a series of 433 free tissue transfers (108 to the lower extremity) in pediatric patients, with a 99.8% success rate and 9% complication rate. Indications for free tissue reconstruction of lower extremity defects in children are similar to those in adults and include avulsion injuries of the distal tibia and foot, injuries involving defects of both soft tissue and bone, extensive soft tissue deficiency, osteomyelitis, failure of other reconstructive techniques, and unstable scars or skin grafts.^{20,21}

Free tissue transfer in pediatric patients offers the advantage of minimal comorbidities, robust wound healing, good sensory/motor regeneration, and the benefit of adaptation and growth of transferred tissue. Pediatric microvascular surgery may require technical modifications based on small vessel size and structural differences of the vessels.^{92,96,97} The walls of the vessels are thinner and more fragile than those of adults, leading some surgeons to recommend the use of systemic anticoagulation in all pediatric free flaps.^{95,97} Larger series, however, have not shown this to be necessary.⁹¹ The absence of atherosclerotic changes in the vasculature of pediatric patients is advantageous to surgeons.⁹⁸ For most cases that do not involve substantial trauma or congenital anomalies, routine preoperative angiography is unnecessary if pulses are palpable.⁹⁹ Duplex ultrasound imaging, MRA, and CTA are less invasive than traditional angiography and may provide anatomic detail information that can help with flap planning and decision-making regarding the recipient site.¹⁰⁰ Other considerations related to microsurgery for pediatric patients include a two-team approach to shorten operative time and careful attention to blood loss. Many surgeons do not give aspirin for postoperative anticoagulation because of concerns about an association between Reye syndrome and aspirin use.¹⁰¹ However, this association has been questioned in recent years.¹⁰²

Many of the massive defects that occur after significant lower extremity trauma require the bulk and/or surface area of rectus and latissimus muscle flaps, which are then covered with split-thickness skin grafts.¹⁰³ The gracilis is an ideal flap for smaller defects in children because of its well-hidden donor scar and minimal morbidity (Fig. 56-17). Muscle flaps are also indicated in cases of osteomyelitis and bony nonunion, because they provide increased blood flow and more complete dead space obliteration.¹⁰⁴ However, in following Gilles's concept of "replace like with like," tissue transferred to reconstruct a wound should be tailored to match recipient site requirements.¹⁰⁵ The dorsum of the foot and the ankle areas are ideally covered with thin, pliable



tissue, such as that provided by skin-grafted fascial flaps or fasciocutaneous flaps from the forearm, thigh (anterolateral thigh), scapular/parascapular, or temporoparietal donor sites.⁹¹ A free-style approach to elevating perforator free flaps has demonstrated early success using an anterolateral thigh perforator, a superficial circumflex iliac artery perforator, an upper medial thigh perforator, and a posterior interosseous perforator.¹⁰⁶ The unique tissue found in the weight-bearing areas of the plantar foot is not entirely replicated by any single tissue type, its ideal replacement is controversial. Muscle flaps such as the rectus abdominis or latissimus dorsi provide bulk to re-create the heel pad, but limited sensation or lack of sensation may lead to breakdown of skin grafts over these flaps.¹⁰⁷ Fasciocutaneous and myocutaneous flaps provide better skin quality and

the possibility of cutaneous sensation, but they are subject to shear and are often bulky, creating problems with footwear.¹⁰⁸

Although reported free flap success rates in the pediatric population are comparable or better to those in adults, postoperative complication rates ranging from 9% to 35% and long-term problems with the reconstructed limb have been reported.^{91,109} Limb discrepancy, valgus and varus deformities, and degenerative arthritis of the ankle are common consequences of the original injury, with bulky flaps, ulceration of skin grafts, contractures, and hypertrophic scars related to flap reconstruction contributing to significant additional functional problems. Wei et al¹⁰⁹ recommended special precautions with the preoperative assessment, planning, and execution of surgery and with the postoperative care of pediatric patients undergoing microvascular lower extremity reconstruction. For example, in patients requiring preservation of shoulder function, they suggested partial latissimus muscle harvest and advocated adequate dorsal foot coverage to prevent contractures and subsequent toe deformities, which can occur as the foot grows.⁹⁵

In general, long-term donor-site morbidity has not yet been well defined in pediatric patients. Donor-site considerations include preventing tight donor-site closures and using fasciocutaneous flaps to facilitate maintenance of donor-site growth characteristics. Particular care is required with the rectus abdominis muscle and radial forearm flap because of their importance for abdominal wall stability and upper limb vascular inflow, respectively. The latissimus dorsi flap should not be used in patients with contralateral shoulder muscle paralysis and in patients who rely ideally on crutches when possible or a wheelchair for locomotion.^{91,92} The psychological consequences of donor-site scars should not be minimized, because visible scars can be the source of significant ongoing distress to children and parents.

Postoperative management of free tissue transfers can be challenging, especially in younger children. Fear and pain are more common in children and can cause flap-compromising vasoconstriction in the early postoperative period. Rinker et al¹³ recommended early visits from social workers and child psychologists to assist with the expected difficulties in adjusting to the injury and hospitalization. They also found that epidural catheters can be very effective in controlling postoperative pain and reducing narcotic use. Sedating patients for procedures such as dressing changes, pin removal, IV changes, and difficult blood draws is essential.⁹²

Protecting the flap in pediatric patients can be problematic. Splinting or casting (with a flap window), trapeze suspension, and triangulated external fixators are useful techniques for maintaining elevation and fixation.¹¹⁰ Positioning must be adjusted carefully to prevent placing pressure on important vascular structures. This is particularly vital when the posterior tibial vessels are used as recipient vessels for distal leg and foot reconstruction. The leg often requires suspension to prevent venous compression and flap congestion.

SKELETAL RECONSTRUCTION

High-energy tibial injuries with combined skeletal and soft tissue injuries should be managed with a combination of orthopedic and plastic surgery approaches.¹⁰⁵ After the initial resuscitation and evaluation, surgeons should determine the extent of injury and the potential for salvage. Most injured extremities can be salvaged using currently available techniques. Patients with severe injuries might be better served with a definitive amputation than with preservation of a nonfunctional, painful limb after limb salvage.¹¹¹ If reconstruction is planned, skeletal stability is obtained. In the treatment of skeletal injuries in adults, immediate internal fixation is preferred for open fractures without bone loss or significant concomitant injury, whereas in children, external fixation is more commonly indicated.¹¹² Severely traumatized lower extremities in patients of all ages often require external fixation.¹⁰⁵ After the soft tissue is reconstructed, persistent skeletal defects can be addressed by bone grafting, bone transport, or free tissue transfer.

Defects up to 5 cm long can usually be reconstructed with conventional nonvascularized bone grafting,¹⁰⁵ but the supply of cancellous bone graft may be limited in smaller children. Complications, including fracture, nonunion, and infection, occur in 10% to 20% of patients undergoing grafting of large bony defects.¹¹³

Posttraumatic tibial defects larger than 5 cm are often managed with distraction osteogenesis, bone transport, or vascularized free tissue transfer. The skeletal defect is reduced with a temporary antibiotic spacer or limb shortening. After the soft tissue heals, the bone is lengthened to fill the bone defect or to restore limb length. Ilizarov¹¹⁴ developed this technique, which has been successfully used in pediatric patients for a variety of skeletal defects.¹¹⁵ Current methods of bone transport allow distraction osteogenesis of 1 to 1.5 mm per day. Reported complication rates range from 5% to 25%.¹¹⁶

Free tissue transfer for reconstructing bony defects of the lower extremity primarily involves the use of a contralateral free fibula transfer.^{109,117} Advantages of free tissue transfer include the ability to restore adequate length with augmentation of local blood supply to the traumatized area. Preserving the bone's vascularity facilitates survival of osteoblasts and osteocytes and healing at the native bone-graft interface without the need for replacement of the graft by creeping substitution.¹¹⁸ The use of vascularized bone may provide sufficient healing potential to reduce the number of procedures required to achieve union, compared with distraction osteogenesis.⁴ In patients with congenital pseudarthrosis of the tibia, this technique has markedly improved the ability to achieve union, compared with conventional bone grafting.¹¹⁹ In addition to these influences on bone healing, the technique enhances growth potential through the inclusion of physes in the flap. The proximal physis contributes 60% of fibular growth and can be transferred with the epiphysis and metaphysis of the fibula using the anterior tibial vessels or peroneal and lateral inferior genicular vessels in combination.¹²⁰ Epiphyseal decancellation can be performed to reshape the flap while preserving the articular surface, periosteum, and physis.¹²¹ Because additional soft tissue can be incorporated in free fibula flaps, they are ideal for managing composite defects.

The smaller caliber of the fibula creates a mismatch in size at the fibula-tibia interface. Controlled loading of the fibula results in remodeling and hypertrophy, but stress fractures occur in 25% to 40% of cases.¹²² The use of a folded free fibula (double- or triple-barrel) increases the cross-sectional area, improving the tolerance of mechanical stress and torque. This technique reduces the risk of fracture and decreases the time to ambulation from 15 months to 6 months.¹²³

Combining free fibula flaps and massive bone allograft has been described for reconstructing bony oncologic defects of the lower limb.¹²⁴ This technique incorporates the mechanical properties of allograft with the biologic properties of vascularized tissue, resulting in earlier stability and incorporation.¹²⁵

TISSUE EXPANSION

Tissue expansion is often used in pediatric patients to manage giant congenital nevi and secondary reconstruction of burn scars.¹²⁶ The technique allows the resurfacing of defects with local tissue of the same color, texture, and thickness. Tissue expansion has a reported complication rate of 9.6%, with expander infection rates ranging from 4.4% to 9.3%, necessitating a low threshold for initiation of antibiotics.¹²⁷ With a reconstructive success rate of 98.3%,¹²⁷ however, tissue expansion has revolutionized the management of large cutaneous defects, anomalies, and scars.

Despite the high success rates associated with tissue expansion, the lower extremity has been found to be an unfavorable location for this technique. Complication rates with expansion in the extremities, especially the lower extremities, are significantly higher than in the scalp, face, and trunk. Problems encountered include expander extrusion, rupture, infection, flap necrosis (both during and after expansion), neurapraxia, and bone resorption.^{127,128} Several theories have been proposed for regional differences in complication rates, including limited tissue availability, geometry of flap design, and problems with vascular supply.¹²⁷

Unacceptably high complication rates are associated with tissue expansion below the knee and in limbs with open wounds.¹²⁸ Current indications for tissue expansion in the lower extremity include correction of contour irregularities, excision of unstable scars or skin grafts, excision of congenital nevi, and secondary correction of burn scars. When tissue expansion is performed in the leg, the following recommendations may minimize the risk of complications^{128,129}:

- In the thigh, incisions for placing an expander should be made at the edge of the defect, whereas at the knee, a remote incision may be less prone to dehiscence.
- A large, single expander should be placed in the subcutaneous plane.
- Expansion should be controlled by patient discomfort and skin blanching. Expansion in this area can take a significant amount of time (from 8 to almost 30 weeks in one series¹²⁹).
- Planned flap advancement should be simple and without a rotational component.
- Complications must be recognized early and treated aggressively.

Additional uses of tissue expansion in the lower extremity include preexpansion of free flaps and expanded, full-thickness skin grafts for lower extremity reconstruction¹²⁷ and preexpansion of dorsal foot skin before clubfoot surgery.⁵

LIMB REPLANTATION

Replantation in the lower extremity can be attempted for pediatric patients with a limb amputation from a sharp or guillotine-type injury with minimal additional soft tissue trauma. Factors most likely to affect outcome include the mechanism of injury, the duration of warm ischemia, and the extent of soft tissue injury.¹³⁰ Unfavorable outcomes are associated with long ischemia time (8 to 10 hours), more proximal injuries, multilevel trauma, and avulsion or crush injuries.^{15,131,132}

Long-term follow-up with children who have undergone limb replantation shows growth of the replanted limbs, although disparity in length between the replanted and unaffected limbs is common.^{131,133,134} Distraction osteogenesis has been successfully used to equalize limb length and to improve rehabilitation.¹³⁴ Insufficient neural regeneration is one of the primary factors affecting the ultimate functional outcome of lower extremity reconstructive efforts. Pediatric patients have an improved ability to heal nerve injuries and have been shown to achieve excellent sensory recovery after leg replantation.^{132,135} Although problems such as limb-length discrepancy, joint contractures, and muscle weakness may prevent completely normal function, the long-term functional outcomes of replanted lower extremities in children are generally very good¹³²⁻¹³⁴ (Fig. 56-18). The reconstructive surgeon should be an integral part of the decision-making process in severe lower extremity trauma. Microsurgical salvage of devascularized tissue can provide a worthwhile outcome.



Fig. 56-18 Incomplete amputation of midfoot connected by small skin island with devascularization of distal part, as scheduled by the trauma team for amputation revision. Consultation with the reconstructive team led to a fairly simple salvage procedure. **A**, Preoperative appearance of incomplete amputation. **B**, Intraoperative appearance after shortening, stabilization, revascularization, and reinnervation. **C**, Lateral view of the late result. **D**, Dorsal view of the late result. **E**, Plantar view of the late result. **F**, Minimal deformity of the revascularized right foot. (Courtesy of Ronald Zuker, MD.)

KEY POINTS

- Lower extremity defects in children are most frequently a result of trauma; however, congenital anomalies and oncologic defects can occur.
- Pediatric patients with lower extremity defects, and their parents, often have a high degree of anxiety about the injury, compounded frequently by the inability to understand and comply with treatment recommendations.
- The potential effects of injury, reconstructive surgery, and donor-site harvest on future growth must be considered when planning treatment.
- Most local, regional, and microvascular techniques used in adults can be successfully applied to pediatric patients.
- Tissue expansion and distraction osteogenesis can be valuable adjunctive methods for reconstructing pediatric lower extremity trauma wounds.

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Evolving Concepts in Pediatric Imaging

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he evolution of modern technology has provided clinicians with numerous sophisticated imaging modalities to aid in diagnosis and treatment planning for surgical conditions. In a relatively short time we have seen an expansion of the imaging armamentarium from simple radiographs to include real-time fluoroscopy and angiography, CT, MRI, MRA, MRV, and more recently, cone-beam CT (CBCT). Advanced imaging modalities have also facilitated virtual planning of complex surgical procedures.

The treatment of pediatric patients with congenital or acquired malformations best exemplifies surgery involving numerous sophisticated imaging modalities. Clinicians should understand the indications, accuracy, advantages, and disadvantages of various imaging modalities. Because concerns over radiation exposure are heightened for children, surgeons should adequately understand these risks and be able to quantify them for concerned parents and decision-makers.

We begin with a brief overview of the imaging modalities commonly employed in pediatric plastic surgery, followed by a discussion of the evidence-based risks of radiation exposure. The rest of the chapter is organized by common conditions treated by pediatric plastic surgeons: craniosynostosis, cleft lip and palate, vascular malformations, and craniofacial trauma. For each condition, we describe how appropriate imaging modalities are employed to guide the workup and surgical treatment. We conclude with a discussion that anticipates future trends in imaging, specifically the growth of virtual surgical planning (VSP) as a heuristic for complex craniofacial procedures.

REVIEW OF TECHNOLOGIES

Radiography

An X-ray is a high-frequency, short-wavelength, high-energy form of electromagnetic radiation that can pass through structures in the body. When directed toward a detector, film or digital, X-rays form a contrasted image based on their differential absorption by tissues such as bone, muscle, fat, and air. Tissues with a higher density and atomic number, such as calcium-rich bone, more readily absorb X-rays and appear white on a radiograph, whereas air-filled structures, such as lung, absorb a minimal amount of X-rays and appear relatively black on a radiograph.¹

Computed Tomography

The English engineer Gregory Hounsfield,² of the eponymous Hounsfield unit, first described his invention of CT scanning in 1972. A CT scanner employs a narrow, fan-shaped beam of radiographs toward a digital detector in a rotational arc around the patient to create a series of cross-sectional images, or axial “slices,” as the patient moves longitudinally through the gantry. Computer algorithms take these cross-sectional slices and generate views in other two-dimensional planes, sagittal and coronal, and in three-dimensional (3D) reconstructions.³ Most modern medical CT scanners use multidetector CT, which uses a large detector arc that acquires up to 64 axial slices simultaneously.⁴ Resolution between axial slices ranges from several mm to as low as 200 μm , depending on machine settings.

CT is optimal for imaging and diagnosing bony abnormalities such as those found in many craniofacial differences. CT images of soft tissues are more difficult, because these tissues absorb X-rays poorly, although visualization can be augmented by the addition of iodine-based contrast agents.

Cone-Beam Computed Tomography

CBCT was first developed in Japan in the early 1990s and became commercially available in the early part of the twentieth century.⁴ Its name derives from the use of a larger, cone-shaped X-ray beam as opposed to the fan-shaped beam employed by traditional CT scanners. CBCT employs smaller and less-expensive components that require less electrical energy than its fan-shaped counterpart.⁵ As a result, CBCT technology allows faster image acquisition at a lower radiation dose and lower cost, compared with traditional CT. Resolution with CBCT is as high as only 70 μm between axial slices; however, in clinical use, accuracy of up to 0.5 mm is possible.⁴ Data collected by a CBCT scanner can be exported to other imaging formats such as panoramic or cephalometric radiographs. Furthermore, CBCT prevents the shortcomings of these traditional imaging modalities, which include insufficient information and a superimposition of anatomic structures.⁵ Limitations of CBCT include poorer soft tissue visualization than with traditional CT.⁴

Micro Computed Tomography

Micro CT scanning technology is capable of producing images of as high as 5 to 50 μm resolution, making it a highly useful tool for research purposes.⁴ However, because it is not employed clinically, it will not be discussed further in this chapter.

Fluoroscopy and Angiography

In contrast to the static images obtained on plain radiographs, fluoroscopy involves continuous X-ray emission through the body to a detector for real-time visualization of the movement of

structures or of the passage of contrast agents through a structure. Angiography involves the use of fluoroscopy after the intravenous injection of a contrast agent to image vascular structures or malformations.⁶

Magnetic Resonance Imaging

MRI involves placing a patient in a magnetic field that aligns the axis of spin of hydrogen nuclei in the body. Hydrogen is used because of its high concentration in water and fat, two of the most abundant tissues in the human body. Radio wave frequency is then pulsed to the field, which causes the hydrogen nuclei to sequentially resonate and relax, emitting a radio wave that is detected by an MRI detector. Various tissues relax at differential rates, resulting in contrast between structures. T1 relaxation refers to the time it takes for the hydrogen nucleus' magnetic vector to return to its original state, whereas T2 relaxation refers to the time it takes for its spin to return to its premagnetization state.⁷

The long-wavelength, low-frequency, low-energy radio waves employed in MRI do not present the same concerns about radiation exposure as do the X-rays employed in CT. MRI is often the optimal imaging modality for examining soft tissue.⁴

Ultrasound Imaging

Ultrasound imaging uses a probe to emit high-frequency sound waves across structures in the body. When these sound waves encounter the interface between two different types of tissue, such as bone and soft tissue, some of the sound waves are reflected back toward and detected by the probe. Using the time to detection of the returning waves, together with the speed of sound, the information from the probe is turned into a two-dimensional image displaying the relative location of structures encountered.⁸

RADIATION EXPOSURE

X-rays are high-energy, ionizing radiation that can excite electrons from their orbits, creating free radicals that cause damage to adjacent DNA molecules.⁹ Concerns about the development of heritable mutations and cancer as a result of radiation exposure are heightened in pediatric patients for several reasons.¹⁰ Epidemiologic studies have shown that children are more sensitive to radiation than adults, probably because children have a higher percentage of rapidly proliferating cells. Furthermore, children have a longer life expectancy than adults, thus they have more time to express the effects of radiation damage.

The effects of radiation vary by tissue type. The cumulative amount of radiation exposure to a patient for a given study is often estimated using the concept of an effective dose unit, which accounts for the measured radiation exposure to each tissue type during a study and weights this exposure based on the radiosensitivity of each tissue type.¹¹ Structures in the head receive a relatively low weight compared with more radiosensitive tissues such as gonadal tissue, breast, and colon.¹¹ However, an effective dose unit is a measure of whole body risk; thus, for the purpose of assessing the risk of a focused examination such as a craniofacial CT scan, reporting of radiation exposure in milliGray (mGy), or the equivalent milliSievert (mSv), is more common.

Jaffurs and Denny¹² analyzed the total radiation exposure of 77 pediatric patients at a single center who underwent thin-slice head or face CT with 3D reconstructions as part of the diagnosis, treatment planning, and postoperative evaluation for either cranial vault remodeling or midface distraction. Nonsyndromic craniosynostosis patients underwent an average of four CT scans, and syndromic craniosynostosis patients underwent an average of 9.73 CT scans over the total course of treatment, with an average radiation dose of 1.39 mSv per CT scan. The total ex-

posure in nonsyndromic and syndromic craniosynostosis patients was roughly 5.6 and 13.5 mSv, respectively. Whether the reported dosages reflect a calculation of brain-specific radiation exposure is not evident in this study. As a frame of reference, each of us receives an effective dose from natural background radiation of about 3.0 mSv each year, half of which is from radon in the soil, although it can be as high as 14.8 mSv for those living at high altitudes.^{12,13}

High-quality evidence on the cancer risk of CT radiation in children has become available in recent years from three international long-term cohort studies. Traditional estimates of radiation risk to children from CT have been extrapolated from the lifespan study of atomic bomb survivors from Nagasaki and Hiroshima, although such extrapolations are fraught with uncertainty and questionable methodology.¹² Inferring individual radiation risk from population-based epidemiologic data is problematic, because it does not account for important factors such as the age at exposure, an individual's genetic predisposition, and particular tissues exposed, which vary significantly in radiosensitivity.¹⁴ These newer studies shed some much-needed light on this important topic.

Most relevant to our patient population is a 2012 study published by Pearce et al¹⁵ in *Lancet*. In a large, retrospective cohort study, the authors found a relative risk of 2.82 for the development of a brain tumor in children who received a cumulative dose of 50 to 74 mGy from head CT, compared with nonirradiated controls. However, in absolute numbers, only 135 of 176,587 (0.076%) patients followed in the entire study developed a brain tumor. The authors estimated that the radiation exposure from head CT accounted for an additional one attributable case of brain tumor per 10,000 head CT scans performed in patients younger than 10 years of age.

Despite low absolute risks, all efforts should be made to minimize the radiation dose to pediatric patients. This is reflected in the As Low As Reasonably Achievable (ALARA) principle advocated by the Society for Pediatric Radiology. Children with craniofacial differences often require multiple interventions and monitoring throughout childhood, thus it is important to remain cognizant of the lifetime attributable risk to these patients. This begins with judicious use of imaging only when indicated. Furthermore, several adjustable parameters affect the total radiation dose for a given study, including tube voltage and current, scan time, and scan field of view.⁵ Surgeons, in collaboration with pediatric radiologists and technical support staff, should ensure proper settings are employed for craniofacial applications in pediatric patients to use the least radiation to provide sufficient image quality. Newer technologies that employ automated tube current modulation automatically adjust tube current using a feedback circuit based on the attenuation value of the previous image, which results in the minimum radiation dosage necessary for sufficient image quality.⁵ CBCT employs significantly lower radiation, and its use should help to meet this goal.

Protective adjuncts, such as leaded eyeglasses and thyroid shields, for particularly radiosensitive organs have been shown to reduce exposure during maxillofacial CBCT.^{16,17} When appropriate indications for ordering examinations are observed and protocols are followed, the benefits from having an accurate diagnosis to guide treatment planning nearly always outweigh risks to the child.

INDICATIONS

Craniosynostosis

CT with 3D reconstruction is a highly accurate imaging modality for evaluating cranial sutures and cranial shape and has replaced plain radiography as the standard for imaging cranial sutures.¹⁸ Nonetheless, whether a CT scan is necessary in all patients with a suspected sutural abnormality

is questionable. For patients with simple, single-suture, nonsyndromic craniosynostosis, experienced clinical assessment is highly accurate, and CT scanning is marginally useful in operative planning or identification of intracranial abnormalities that will change management.^{18,19} Although associated abnormalities such as ventriculomegaly and prominent extra-axial CSF are common, incidental findings that require intervention are rare.²⁰ Furthermore, the traditional study of a plain radiograph remains cost effective and a useful adjunct in patients with a low pretest probability of craniosynostosis based on clinical examination.²¹ Thus routine CT imaging is not required, and the benefits may not outweigh the combination of radiation risk, the possible need for sedation to obtain an adequate study, and resource utilization.

In contrast, 3D CT is a powerful tool for preoperative assessment and planning for syndromic craniosynostosis (Fig. 57-1). Traditional skull radiographs have a low sensitivity to detect complex

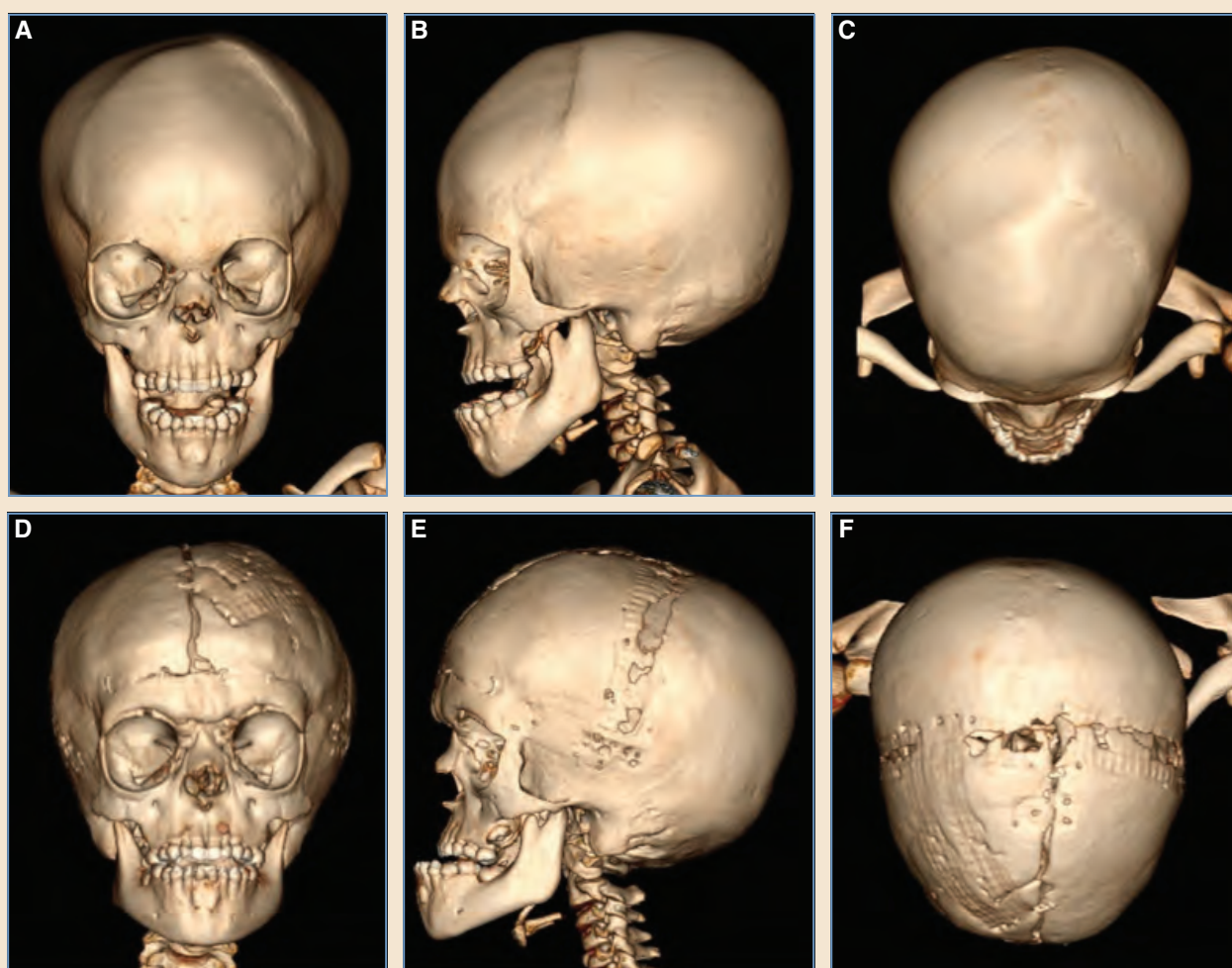


Fig. 57-1 A-C, 3D reconstructions of a low-dose craniofacial CT scan obtained to aid diagnosis and preoperative planning for a 6-year-old male with Apert syndrome. The patient presented with pansutural craniosynostosis, resulting in papilledema and thumbprinting of the calvaria, suggestive of elevated intracranial pressure. D-F, 3D reconstructions of a low-dose craniofacial CT scan obtained 1 year after anterior cranial vault remodeling, frontoorbital advancement, and frontal sinus obliteration. The patient also had midface hypoplasia and shallow orbits characteristic of Apert syndrome.

and minor sutural craniosynostosis.²¹ Low-dose spiral multidetector CT with 3D reconstruction allows not only an identification of major sutures involved in multisuture craniosynostosis, but also characterizes the involvement of minor skull-base sutures and facilitates an accurate characterization of supraorbital rim deformities and nasal root deviation.²² CT is also useful for evaluating brain structural abnormalities that have a higher degree of association with syndromic or multisuture craniosynostosis, such as hydrocephalus, ventricular dilation, brain hemiatrophy, chronic subdural hematoma, and agenesis of the corpus callosum.²¹

Routine postoperative screening CT is not recommended for patients undergoing cranial vault remodeling. A review of patients undergoing routine postoperative CT showed no change in management or prognosis based on the scan, with the exception of patients with concomitant shunt placement for hydrocephalus.²³ However, an understanding of changes in cranial vault volume over time until skeletal maturity is highly useful. For this purpose, a high degree of correlation has been shown between the use of 3D photography and 3D CT, and some authors have advocated this as an alternative to track cranial development over time without the risks of radiation exposure.²⁴

Multiple authors have investigated the utility of ultrasound imaging as an alternative to CT for diagnosing single- and multiple-suture craniosynostosis and have found it to be a highly sensitive and specific alternative to CT.²⁵⁻²⁷ Given its high negative predictive value, ultrasound scanning has been suggested as an initial screen in suspected infants, and CT is recommended for cases in which ultrasonography results are abnormal or equivocal or when needed for preoperative planning.²⁸ A potential drawback of ultrasound imaging is its operator dependence; thus experienced technicians help to increase accuracy.

The use of MRI with novel parameters to optimize visualization of the soft tissue–bone interface has also been reported, allowing 3D cranial reconstruction and accurate diagnosis of craniosynostosis.²⁹ However, the potential need for lengthy sedation or general anesthesia is a drawback for this modality and a reason it is not widely used.

CBCT offers substantial promise to produce high-quality 3D reconstructions for planning and postoperative evaluation of craniosynostosis patients, with a fraction of the radiation dose and cost of traditional CT scan. However, it is not widely used for this indication. This indication for CBCT is discussed further in the section on VSP.

Cleft Lip and Palate

In patients with cleft lip and palate, several indications exist for multiyear imaging and for using a comprehensive team approach. Assessment of velopharyngeal function after cleft palate repair is important as these children age, because evidence of velopharyngeal inadequacy may necessitate a secondary palatal procedure. Evaluation by a trained speech and language pathologist, multiview videofluoroscopy, and video nasopharyngeal endoscopy are important adjuncts for assessment.³⁰ Multiview videofluoroscopy allows real-time visualization of the velopharyngeal mechanism in multiple views. A contrast material is injected into a patient's nares and fluoroscopic images are recorded for later review. The child is directed to produce certain patterns of speech. During nasal endoscopy, patients are instructed to repeat key phrases that stimulate velopharyngeal closure, while a small, flexible endoscope views the lateral and posterior pharyngeal walls and the nasal surface of the velum. Both modalities provide useful information, although the velopharyngeal closure pattern is better assessed with nasendoscopy and may be more helpful in guiding further surgical intervention.^{30,31}

The next milestone in the treatment of patients with cleft lip and palate is often alveolar bone grafting. Traditionally, two-dimensional dental radiographs have been used to guide decision-making for this procedure. However, CT has proved superior to standard radiographs to characterize the alveolar defect volume preoperatively and the bone graft volume and resorption postoperatively.³² More recently, CBCT has emerged as the preferred imaging modality for this purpose given its decreased radiation dose, lower cost, and ease of use (Fig. 57-2).³³ CBCT has been shown to be highly accurate and precise for this indication when measured against a water-displacement method in an experimental model.³⁴ The CBCT field of view can be extended to evaluate calvarial bone as a potential donor site for maxillary grafting.³⁵ Furthermore, CBCT can be used to plan and evaluate osteointegrated dental implants after alveolar bone grafting; oral surgeons already use it routinely for this purpose.³⁶ CBCT scans should be reviewed by multiple members of a multidisciplinary team, because incidental findings of dental, airway, sinus, and ear abnormalities are common in these patients.³⁷

A subset of cleft palate patients will eventually require midface advancement. Although traditional cephalograms have been the mainstay of orthognathic planning, more recently, CT and CBCT have been used for quantitative 3D preoperative planning for midface advancement in adolescents with cleft lip and palate.³⁸

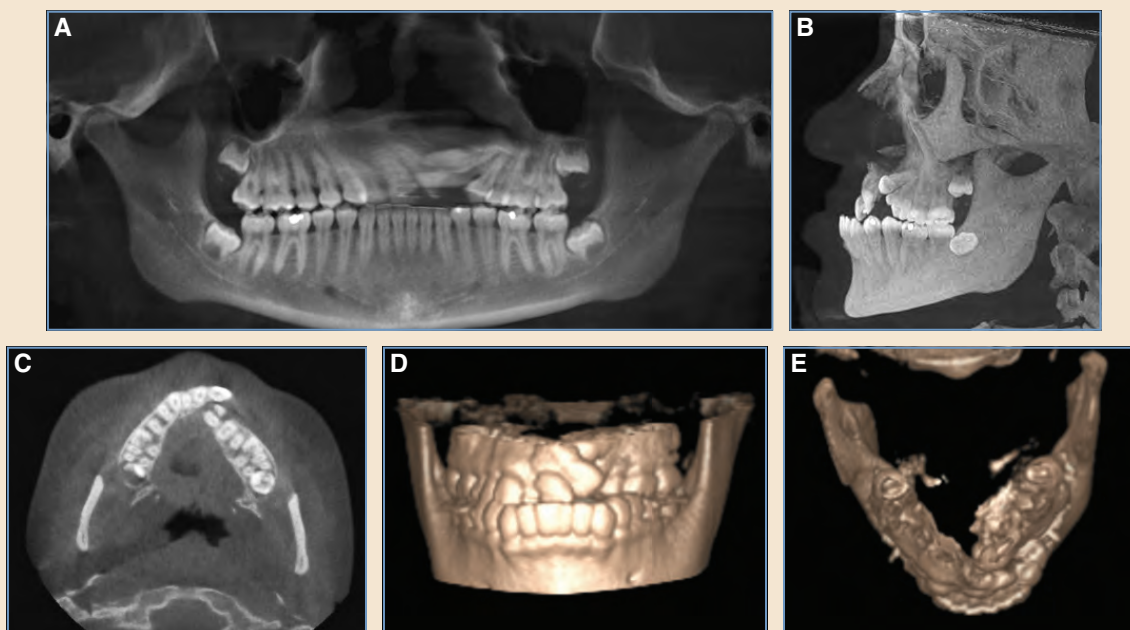


Fig. 57-2 CBCT scans of a 15-year-old female with a history of left cleft lip and palate. The scans were obtained for a preoperative workup before palatal expansion and alveolar bone grafting. The variety of image formats demonstrates the versatility and utility of CBCT. **A**, A traditional Panorex image generated from the CBCT scanner. **B**, A lateral cephalogram generated from the CBCT scanner, highlighting the patient's midface hypoplasia and negative overjet. **C**, An axial CBCT cut demonstrates well the patient's left-sided palatal collapse and alveolar bone deficit. The patient will require palatal expansion to reestablish the proper position of the maxillary arch, followed by alveolar bone grafting of the bony defect to prepare for an osteointegrated dental implant. **D** and **E**, An AP and worm's-eye view of 3D reconstructions generated using CBCT data.

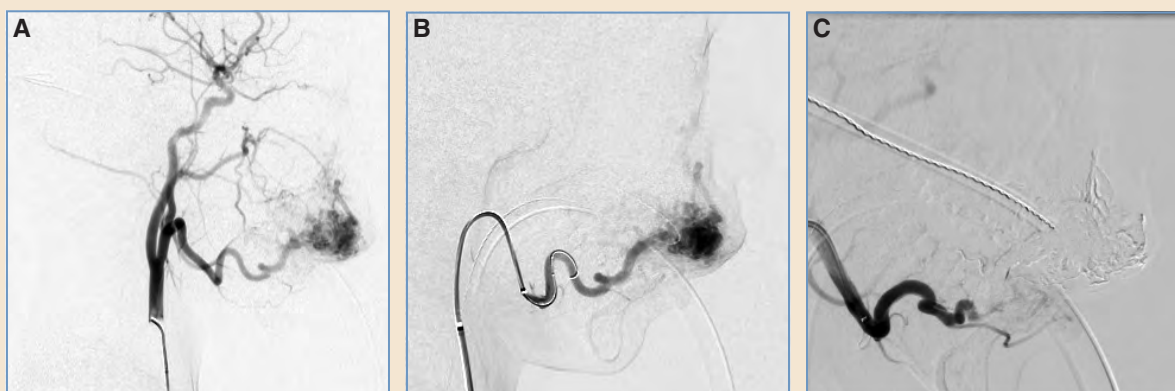


Fig. 57-3 An arteriogram obtained during the preoperative embolization of an arteriovenous malformation (AVM) of the right face and right upper lip in an 11-year-old female. The AVM was first noted when the child was young and continued to enlarge over time. She underwent her first surgical resection (with incomplete excision) at 7 years of age at a different institution. **A**, A right common carotid artery angiogram showed blood supply to the AVM from the right facial artery. Fluoroscopy also clearly demonstrated venous drainage through the external jugular and retrograde through the angular vein. Such precise delineation of the vascular anatomy of a malformation can be very valuable information to guide operative planning and execution. **B**, An intraprocedure angiogram showed positioning of a sceptor balloon catheter within the distal right facial artery before embolization with Onyx. **C**, An angiogram performed after the embolization showed no blood flow to the lesion through the facial artery. Embolization achieved 100% occlusion of the AVM. The patient underwent excision of the vascular malformation with local tissue rearrangement for wound closure.

Vascular Anomalies

Vascular anomalies typically are diagnosed clinically; however, imaging is a useful adjunct in difficult diagnostic cases and for surgical planning. Multiple modalities, including ultrasonography, CT, CTA, MRI, MRA/MRV, and traditional fluoroscopic angiography, can be used to image vascular anomalies, depending on the location, the type of lesion, and preoperative planning needs.

Ultrasound imaging is often performed first, because it is free of radiation and does not require sedation. When combined with color Doppler ultrasonography, ultrasound imaging can provide information not only on structure but flow within a lesion.³⁹ However, MRI remains the standard for imaging vascular lesions given its superior ability to delineate soft tissues and fat signal suppression to clearly demarcate the anatomic boundaries of a lesion.⁴⁰ The use of MRA/MRV can help to identify a feeding artery and draining vein, similar to a traditional arteriogram and venogram, which can be important for surgical planning (Fig. 57-3). However, CT with contrast may prove more useful for lesions with osseous involvement.⁴⁰ Working closely with a pediatric radiologist will help surgeons to choose the optimal study.

Facial Trauma

Maxillofacial CT is the standard imaging modality for assessing pediatric patients with suspected facial fractures based on the history and clinical examination. The radiation dosage should be

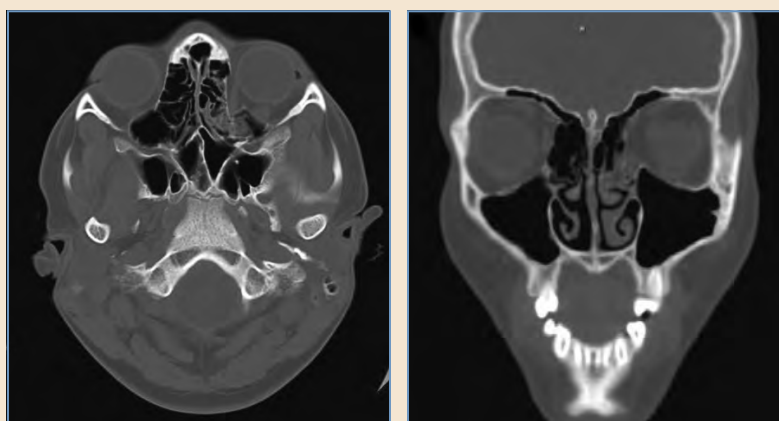


Fig. 57-4 Axial and coronal CT slices of the facial bones show an entrapped medial orbital wall fracture. This 12-year-old male was kicked in the face during a martial arts class. He presented with slight restriction of superolateral gaze of his left eye with associated severe pain and diplopia. Mild bradycardia occurred with extreme lateral gaze. The patient underwent an exploration of the medial wall fracture through a transconjunctival incision. The medial rectus and orbital contents were reduced, and a resorbable plate was placed to repair the medial orbital wall.

kept as low as possible, although good data on the sensitivity of low-dose CT scan protocols for pediatric facial trauma are not available.⁴¹ Panoramic radiographs have some utility in evaluating mandibular fractures; however, interpreting plain radiographs in children is complicated by underdeveloped sinuses and tooth buds that can obscure fracture lines.⁴² Additional drawbacks of panoramic radiographs in patients with facial trauma include the need for patients to be motionless to obtain an adequate image, and, in those with spinal precautions who cannot be placed in the required supine position, it often cannot be performed.⁴³ CT scanning can help to detect subtle fractures of the mandibular condyle not seen on AP radiographs or panoramic radiographs, and it is also a sensitive modality for detecting fractures of the medial orbital wall and orbital floor⁴ (Fig. 57-4). 3D reconstructions can facilitate visualization of fracture patterns and surgical planning.

VIRTUAL SURGICAL PLANNING

CT-guided VSP is the newest and potentially most exciting evolving concept in pediatric imaging. It offers the opportunity for improved accuracy and decreased operative times for novice surgeons and can be a valuable tool for even the most seasoned surgeons in planning complex craniofacial reconstructions. Fig. 57-5 shows the use of VSP to correct a complex deformity after an earlier LeFort III midface advancement and subsequent counterclockwise rotation of the LeFort III segment. VSP was used to plan clockwise derotation of the LeFort III segment. Measurements for the planned movements at multiple reference points on the facial skeleton were determined through VSP (Table 57-1), and demonstrated on cephalometric projections. A LeFort I osteotomy will be performed at a later date to correct the occlusion.

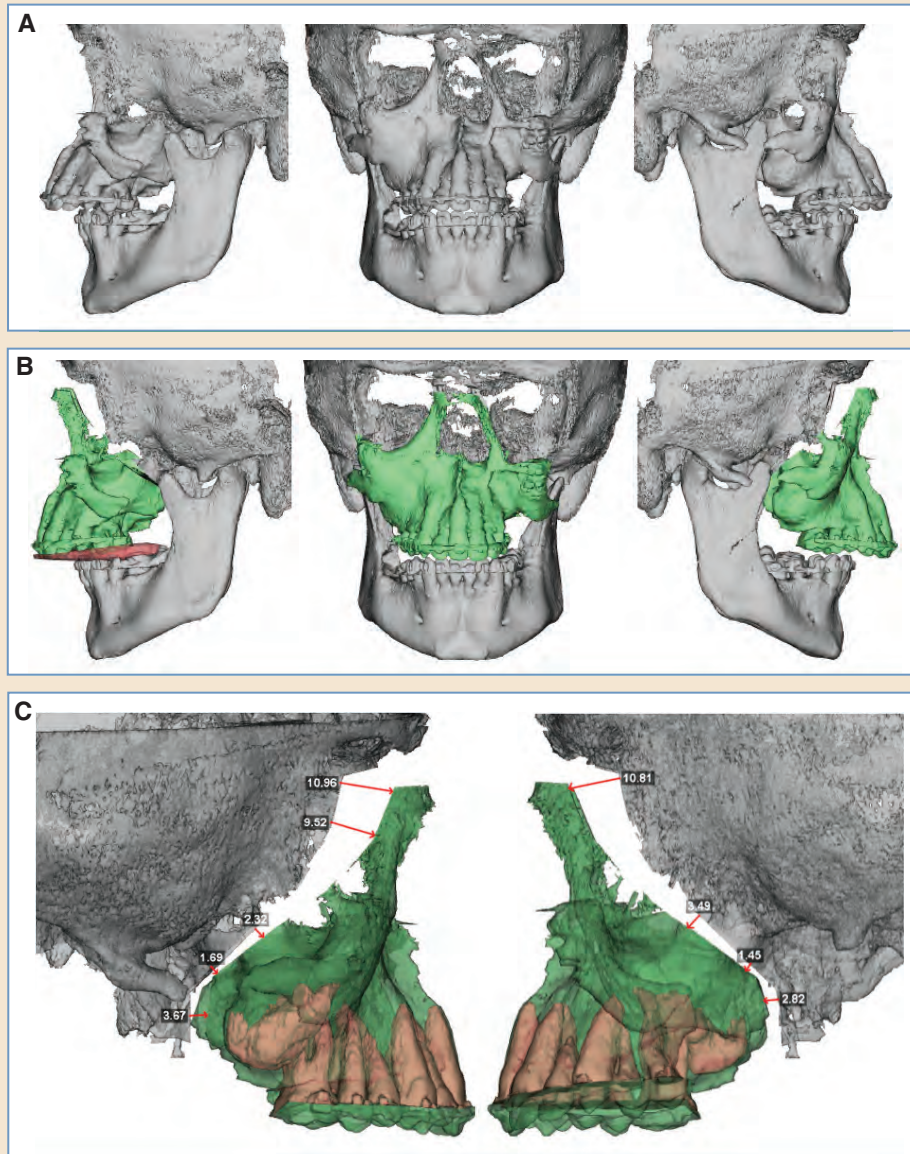


Fig. 57-5 VSP to correct a complex craniofacial deformity after a LeFort III midface advancement. **A**, 3D CT images show counterclockwise rotation of a LeFort III segment. **B**, VSP was performed to plan a clockwise derotation of the LeFort III segment. **C**, Cephalometric projections from the planned movement are demonstrated.

Table 57-1 Measurements for Planned Movements at Multiple Reference Points on the Facial Skeleton as Determined Through Virtual Surgical Planning

Point	Name	Anterior or Posterior	Left/Right	Up/Down
ANS	Anterior nasal spine	5.56 mm anterior	0.00	3.47 mm down
A	A point	4.46 mm anterior	0.00	3.56 mm down
ISU1	Midline of upper incisor	0.19 mm posterior	0.00	4.82 mm down
U3L	Upper left canine	0.05 mm posterior	0.00	2.58 mm down
U6L	Upper left anterior molar (mesiobuccal cusp)	0.17 mm anterior	0.00	0.09 mm down
U3R	Upper right canine	0.08 mm posterior	0.00	3.87 mm down
U6R	Upper right anterior molar (mesiobuccal cusp)	0.10 mm anterior	0.00	0.90 mm down
ISL1	Midline of lower incisor	2.62 mm posterior	0.06 mm left	4.13 mm down
L6L	Lower left anterior molar (mesiobuccal cusp)	2.43 mm posterior	0.07 mm left	1.97 mm down
L6R	Lower right anterior molar (mesiobuccal cusp)	2.81 mm posterior	0.08 mm left	2.27 mm down
B	B point	3.97 mm posterior	0.11 mm left	3.86 mm down
Pog.	Pogonion	4.93 mm posterior	0.14 mm left	4.02 mm down

Herlin et al⁴⁴ reported on the use of VSP for preoperative volumetric measurements and simulation, for design of cranial bone grafts, and for fabricating intraoperative cutting guides to treat patients with Treacher Collins syndrome. Shah et al⁴⁵ argued that the use of 3D CT scanning for preoperative simulation of frontoorbital advancement and fabrication of intraoperative cutting guides to be used for supraorbital bandeau osteotomies and manipulation can lead to more precise and reliable results in patients with metopic craniosynostosis. Furthermore, preoperatively planned frontoorbital advancements can be accurately achieved intraoperatively.⁴⁶ A recent multiinstitutional study has demonstrated an improved conformity of the operated supraorbital bandeau with ideal normative shape and a significant decrease in operative time using VSP and prefabricated templates to treat patients with metopic craniosynostosis.⁴⁷

CBCT also shows much promise for use in VSP. Measurements for LeFort I osteotomies using both traditional CT and CBCT have been shown to correlate well with each other and with direct physical measurement.⁴⁸ Katkar et al⁴⁹ showed high interobserver and intraobserver reliability for identification of cephalometric landmarks for orthognathic surgical planning using two of the most commonly used commercially available CBCT machines—Galileos (Sirona Dental Systems) and Next Generation iCAT (Imaging Sciences International)—in combination with Dolphin 3D software. Some authors have combined the use of traditional plaster models with CBCT-based VSP for orthognathic planning.⁵⁰ Facility with the use of VSP software and integration of these technologies into the preoperative planning of complex craniofacial and orthognathic procedures will increasingly become the norm for the coming generation of craniofacial surgeons.

KEY POINTS

- Clinicians must understand the indications, accuracy, advantages, and disadvantages of various imaging modalities to diagnose and treat pediatric patients with congenital or acquired malformations.
- CT is optimal for diagnosing bony abnormalities. CT visualization of soft tissues can be augmented by the addition of iodine-based contrast agents.
- CBCT allows faster image acquisition, with a lower radiation dose and lower cost, compared with traditional CT.
- MRI is often the optimal imaging modality for examining soft tissue.
- Despite low absolute risks, all efforts should be made to minimize the radiation dose to pediatric patients, including judicious use of imaging only when indicated and employment of proper equipment settings for pediatric craniofacial applications.
- For assessing patients with simple, single-suture, nonsyndromic craniosynostosis, CT imaging is not required, and the benefits may not outweigh risks. In contrast, 3D CT is a powerful tool for preoperative planning for syndromic craniosynostosis. Routine postoperative screening CT is not recommended for cranial vault remodeling.
- Multiview videofluoroscopy and video nasopharyngeal endoscopy are important adjuncts in assessing children with cleft lip and palate for velopharyngeal inadequacy. CBCT has emerged as the preferred imaging modality to evaluate these patients for alveolar bone grafting.
- Multiple modalities, including ultrasound, CT scan, CTA, MRI, MRA/MRV, or traditional fluoroscopic angiography can be used to image vascular anomalies, depending on the location, the type of lesion, and preoperative planning needs.
- Maxillofacial CT is the standard imaging modality for assessing pediatric patients with suspected facial fractures based on history and clinical examination.
- CT-guided VSP is the newest and potentially most exciting evolving concept in pediatric imaging, offering the opportunity for improved accuracy and decreased operative times for novice surgeons. It can also be valuable for more seasoned surgeons in planning complex craniofacial reconstructions.

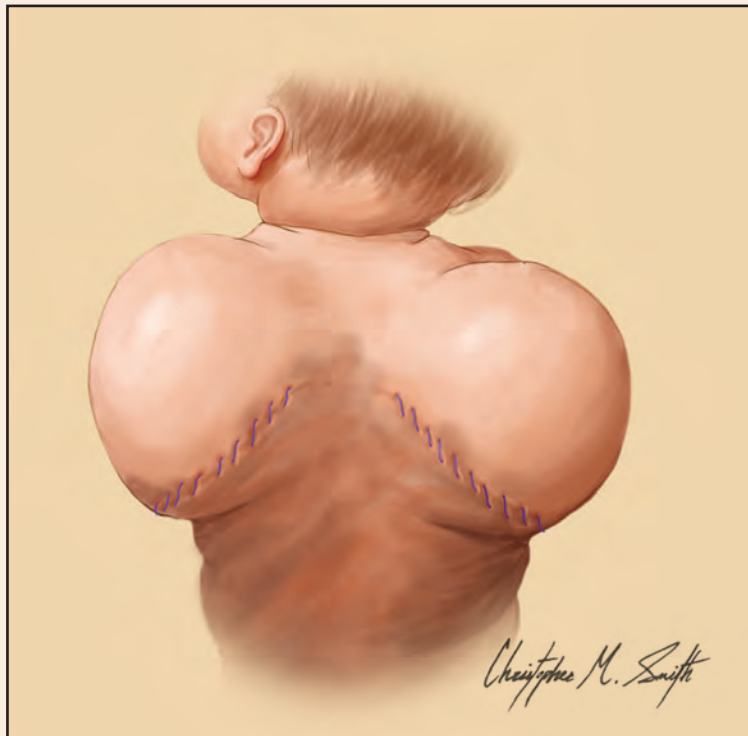
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Part IV



Congenital and Acquired Deformities of the Thorax, Abdomen, and Back

58

Congenital Deformities of the Thorax and Chest Wall

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Nuss Repair of Pectus Excavatum.



ongenital thoracic deformities consist of a handful of bony and cartilaginous defects that are usually present at birth and may persist into adulthood without intervention. The most common congenital thoracic deformities include Poland syndrome, pectus excavatum, pectus carinatum, and less frequently, Jeune syndrome and pentalogy of Cantrell. Although chest wall deformities in children were described as early as the fifteenth century, operative management of these defects did not bloom until the twentieth century.¹ Today, the considerations, techniques, and controversies regarding the management of these complex and difficult deformities are numerous. Most of the foundational thoracic, pediatric, and plastic surgery literature is still applicable today; however, some newer and evolving approaches exist.

POLAND SYNDROME

Poland syndrome is characterized by chest wall and upper extremity defects. Unilateral chest wall defects include partial or complete absence of the pectoralis major and minor muscles and their associated subcutaneous fat, hypoplasia of the ipsilateral breast or nipple, absence of axillary hair, absence of the natural anterior axillary border, and anomalies of the costal cartilages and anterior rib ends. Associated upper extremity defects include limb shortening, syndactyly and/or brachysyndactyly, carpal anomalies,² and hypoplasia/aplasia of the middle phalanges, which multiple authors have classified.^{3,4} Less frequently, patients have deformities of the serratus, infraspinatus,

latissimus dorsi, and external oblique muscles; dextrocardia⁵; or complete absence of the hand. The syndrome may also occur with congenital bilateral facial paralysis and the inability to abduct the globe, referred to as *Moebius syndrome*.⁶ Some patients have an associated hematopoietic malignancy or vertebral abnormalities. Although most cases are unilateral, bilateral symptoms are occasionally reported in the literature, with a widely variable phenotypic spectrum.⁷

History and Epidemiology

Poland,⁸ an English medical student, first described Poland syndrome in 1841, during the post-mortem evaluation of a convicted murderer. He noted that the patient had thoracic wall deformities, including a defect of the pectoralis major, confined to the musculature on one side of the chest.⁹ He also noted that in one hand the middle phalanges were absent in all fingers except the middle finger, and that the hand had webbing between the proximal phalanges and was shorter than the opposite hand. Previously, Lallemand in 1826 and Froriep in 1839 had described the absence of the pectoralis muscle without associated hand deformities.¹⁰ Poland syndrome may be underreported, because it is often undiagnosed. The prevalence is approximately 1 in 30,000 births. Estimating sex differences is difficult, because more women are presumed to present to surgeons for breast deformity correction. The deformity occurs most often on the right side.

Genetic and Etiologic Factors

The cause of Poland syndrome is unknown and controversial. Possible causes include an abnormal migration of embryonic tissues forming the pectoral muscles; hypoplasia of the subclavian, vertebral, and internal thoracic arteries; pharmacologic use during pregnancy; and in utero injury.¹¹⁻¹³ Poland⁸ noted during his landmark dissection that the internal thoracic arteries were smaller on the affected side. The vascular interruption theory is supported by studies of fetal vasculature development and the possible correlation between known structural abnormalities, including Moebius syndrome, Klippel-Feil syndrome, and Sprengel deformity. The hypothesis is that an insult to the developing vascular tree at a specific point in time¹⁴ during development of the pectoralis major sternocostal head, separation of the fingers, and chondrification of the middle phalanges³ will lead to predictable structural defects. Genetically, most cases appear to be sporadic; only a small number of reported genetic transmissions are consistent with an autosomal dominant gene, low penetrance, and a delayed mutation.^{11,13,15-18}

Functional Implications

The spectrum of chest wall abnormalities includes the following¹⁹:

- A normal thoracic cage with only pectoralis muscle absence
- Ipsilateral thoracic cage and sternal depression with or without contralateral sternal protrusion and rotation
- Ipsilateral rib hypoplasia without significant depression or protrusion of either the thoracic cage or the sternum
- Rib aplasia with depression of adjacent ribs and sternal rotation

Although all patients with Poland syndrome have absence or hypoplasia of the pectoral muscles, not all patients have bony abnormalities severe enough to require surgery. Whether the syndrome defects can be severe enough to compromise the cardiovascular system is controversial. Most cases are postpubescent females who present for cosmetic correction; however, some adolescents and males present with rib aplasia and resultant lung herniation or severe sternal depression that has caused thoracic constriction.

Cosmetic Implications

Most patients seen by plastic and thoracic surgeons are postpubescent females with rib abnormalities and breast hypoplasia/aplasia. Some surgeons advocate waiting to perform reconstruction until after breast maturity.²⁰ Definitive treatment of pubertal females is more difficult and must be individualized based on the patient's and parents' concerns.²¹

History and Assessment

Many patients with absent ribs are candidates for reconstruction, given the potential for lung herniation or susceptibility to thoracic trauma. Female patients merit intervention regardless of the functional considerations, because they invariably have cosmetic deformities by the time they seek surgical evaluation. Males can also have a significant cosmetic deformity. The deformity should be defined with CT imaging, preferably three-dimensional scanning. MRI can be helpful for further definition of the anatomy. These images are useful not only for defining the deformity, but also for indentifying surrounding anatomy for reconstructive options.

Treatment Options

Reconstructive considerations are focused on repairing the underlying bony structure, creating a symmetrical soft tissue structure, and creating an aesthetic breast (which may necessitate a mastopexy of the contralateral side). It has been shown that each of these reconstructive steps can be safely performed in a single operation.²² The bony structural layer is generally reconstructed by replacing the aplastic ribs on the affected side with contralateral rib grafts. The grafts can be split in half and used separately to replace multiple rib levels. Surgeons should not attempt to "build up the chest wall" with implant prostheses alone, because this does not correct the underlying structural defect. A sternal osteotomy and rotation may also be required, because the sternum can often be rotated toward the defect. Once the ribs are stabilized, they are covered with a synthetic graft to create a more natural contour. However, an autologous muscle flap achieves the same purpose without the potential complications of synthetic materials and may provide more bulk (particularly if innervation is maintained) and aid in the breast reconstruction. More recently, autologous fat grafting has been described for volume addition and contouring.^{23,24}

The latissimus dorsi muscle flap, supplied by the thoracodorsal vessels and nerve, is the primary flap used to reconstruct the chest wall in patients with Poland syndrome. This muscle provides sufficient tissue to improve symmetry with the opposite side and has been described to restore function as well.²⁵ A limited dorsal thoracic incision is made to mobilize the muscle from its origin. A second incision is made in the axilla to identify the vascular pedicle and to detach the muscle from its humeral insertion. The latissimus dorsi muscle is then transposed to simulate the deficient pectoralis muscle, to create an anterior axillary fold, and to cover the breast implant if a prosthesis is used or anticipated. Rarely, a patient has an associated ipsilateral latissimus muscle aplasia, and microsurgical transposition of the contralateral muscle may be performed.²⁶

Reconstruction options for the breast improved significantly with the advent of breast implant prostheses, particularly silicone and Silastic (Dow Corning) implants. Once the latissimus muscle is transposed, a prosthesis is inserted through the axillary incision. Some have advocated tissue expansion earlier in childhood, with expansion paralleling the growth of the contralateral breast.^{20,27,28} The expander is replaced with a permanent implant at maturity.

Japanese surgeons Ohmori and Takada²⁹ described their technique of reconstruction with and without silicone prostheses, associated with a myocutaneous flap. Two years later, Hester and

Bostwick²¹ described reconstruction for 11 patients using a latissimus dorsi muscle transposition with and without implants. This reconstruction has been modified by twisting the muscle flap between its transposed origin (between the deltoid and the biceps) and its insertion, a technique that creates a fuller anterior axillary fold.³⁰ Although some surgeons believe mild to moderate deformities can be reconstructed without skeletal reconstruction, most agree that it makes reconstruction difficult and that aplastic ribs, in particular, should first be corrected.^{31,32}

Surgical Technique

An incision is placed along the anterior border of the latissimus, or in female patients, horizontally in the bra line, depending on the patient's preference and required chest wall access. Skin is then elevated off the latissimus dorsi, and medial, lateral, and inferior attachments of the muscle are divided. The humeral attachment is divided, completely freeing the muscle as an island on its neurovascular pedicle. A tunnel is dissected anteriorly to connect with an anterior chest pocket that is dissected through an inferolateral submammary incision. The muscle is then passed through the tunnel and placed in the anatomic space normally occupied by the pectoralis muscle. Inferiorly, the latissimus is sutured to the chest wall fascia at the level of the inframammary crease. Medially, the muscle is sutured to fascia overlying the lateral sternum, and superiorly, it is sutured to the clavicular portion of the pectoralis major. The anterior axillary fold is created by suturing the detached insertion of the latissimus dorsi to the periosteum of the humerus, just below the insertion of the clavicular head of the pectoralis major. After an implant is placed under the latissimus, sutures are secured laterally to prevent lateral displacement. Care is required to ensure that blood flow to the vascular pedicle is not compromised.

Postoperative Care

Patients are instructed not to participate in contact sports for 6 months.

Treatment Outcomes and Complications

Long-term results are excellent with the use of the reconstructive techniques described previously and autologous tissue transfer. Major complications include damage to the latissimus dorsi neurovascular bundle and pneumothorax. An intact nerve maintains muscle bulk but can be associated with muscle overactivity. Deliberate division of the thoracodorsal nerve is controversial.

PECTUS EXCAVATUM

Pectus excavatum (funnel chest), a concavity of the anterior chest wall, is the most common congenital chest wall deformity. This variable deformity is a result of abnormal costal cartilage position and growth, leading to posterior angulation of the cartilages below the manubrium, posterior curvature and displacement of the sternum, and a diminution of the AP diameter of the thoracic cage (Fig. 58-1).

History and Epidemiology

Pectus excavatum was first documented in 1594. In 1949 Ravitch³³ from Johns Hopkins University best described the condition. He noted the characteristic sternal depression, rounded and sloped shoulders, a slight dorsal kyphosis, a prominent potbelly, and retraction of the sternum on deep inspiration. The association with cardiopulmonary dysfunction is controversial. Other associations include Marfan syndrome, scoliosis, hypomastia, cardiac murmurs, and ECG changes.³⁴



Fig. 58-1 Pectus excavatum in an adolescent.

Most cases are identified in early infancy. The depression can become more pronounced as patients grow, although some resolve. The male/female ratio is 3:1.

Genetic and Etiologic Factors

The most plausible cause of pectus excavatum is overgrowth of abnormal costal cartilage, which depresses the sternum in an often-rotated position. The initial theory of pectus excavatum, supported by multiple authors, was that of a shortened substernal tendon pulling the sternum posteriorly during growth.^{35,36} Ochsner and DeBakey³⁷ discounted this theory in 1939, proposing their “abnormal cartilage” theory. Feng et al³⁸ have substantiated this theory in their biomechanical, morphologic, and histologic review,³⁸ although it has been challenged.^{39,40} However, the cartilage theory describes only the gross mechanical physiology. A number of pathogenetic hypotheses have been suggested, but the exact cause leading to the deformity is not known. A positive family history has been noted in up to 43% of cases.^{41,42}

Preoperative Assessment

Care is most often sought by the adult patient (or by parents for their young child) for cosmetic concerns, although some patients may have symptomatic respiratory problems. The effect of a chest concavity deformity on the cardiopulmonary system is controversial, as shown in detailed reviews.^{43,44} Although it seems intuitive that compression of the mediastinum by a depressed sternum would lead to cardiovascular and pulmonary impairment, both anecdotal and scientific evidence have failed to concretely show improvement in status after surgical repair.⁴⁵⁻⁴⁷ Recent studies have shown that deterioration in pulmonary function may be present after operative correction.^{44,47,48} More recently, Malek et al⁴⁹ conducted a meta-analysis of eight studies meeting strict inclusion criteria and that suggested cardiovascular function improved. These results have been challenged.^{50,51} Nonetheless, several studies indicate a significant improvement in body image, perceived ability for physical activity, and subjective patient satisfaction.^{46,52-55} The basis for such controversy is most likely a result of the complex measures used to evaluate cardiac and pulmonary function, the variability in parameters between individuals,⁴⁰ and the variability

in study designs.⁴⁷ Preoperative cardiopulmonary testing, such as pulmonary function tests and ECG, may be useful in patients with severe mediastinal compression.

Complex radiographic studies are not needed for a pectus repair itself, although CT scans have been used as an indication guideline in some series.⁵⁶ Appropriate consultation should be sought in patients with associated anomalies, including Marfan syndrome and scoliosis. Metal allergy ranges from 2% to 15% and may warrant preoperative testing if the use of titanium constructs is considered, as in a Nuss procedure.^{54,57} The patient age for pectus reconstruction is based on each case, although the age at operation appears to be trending upward nationally.⁵⁸ The reasons for performing early surgery include technical ease and less complex reconstruction. However, because the operation is performed for cosmetic appearance in most cases, some argue that the child should make this decision when older. Moreover, after a reconstruction, the chest wall deformity can recur as a child grows.

Treatment Options

The initial surgical treatment for pectus excavatum in the early 1900s consisted of complete resection of the deformed ribs and sternum, which led to paradoxical breathing, an unprotected heart, and a poor cosmetic result.⁵⁹ Subsequently, resection of only the rib ends with elevation of the sternum evolved. In this early technique, traction sutures were placed through the skin and suspended above the patient's bed. Ochsner and DeBakey³⁷ pioneered the common technique of cartilage resection with sternal support. Similar to earlier methods of fixation, they used a metal band around the sternum that was connected to rubber bands attached to an external plaster cast.

Currently, three major options are available to patients with pectus excavatum:

1. Soft tissue or prosthetic augmentation
2. Both open sternal or costal cartilage manipulation and resection with internal or external sternal support (see Fig. 58-2)
3. The innovative minimally invasive technique devised by Nuss et al,⁵⁹ which requires neither a cartilage incision or resection

Soft Tissue or Prosthetic Augmentation

Soft tissue and prosthetic reconstruction involve camouflaging the defect with a silicone or Silastic implant. Initially, these implants were placed subcutaneously, but they are now placed submuscularly to prevent anterior displacement.⁶⁰⁻⁶³ They may also be fixated with a synthetic patch. This option is best for patients who are fully grown and have no subjective physiologic concerns (the previously mentioned cardiopulmonary concerns).

Open Sternal or Costal Cartilage Manipulation and Resection With Internal or External Sternal Support (Ravitch Procedure)

Open sternal reconstructions are generally modeled after the Ravitch procedure, which includes excision of costal cartilages in a subperichondrial plane for the full length of the deformity, separation of the intercostal bundles from the sternum, and an anterior sternal osteotomy for elevation without external fixation.³³ The modified Ravitch procedure involves removal of three to four bilateral overgrown cartilages, with anterior repositioning of the sternum through a posterior transverse osteotomy with internal fixation or support. The sternum is predominantly supported by the lower ribs (Fig. 58-2).

Generally, a temporary support bar or wire is inserted behind or through the sternum for support. Potential modifications to this procedure are numerous, and nearly all employ bilateral resection of abnormal cartilage, a sternal osteotomy, and sternal fixation in the corrected position. They generally differ with respect to the method of sternal fixation. Examples of these

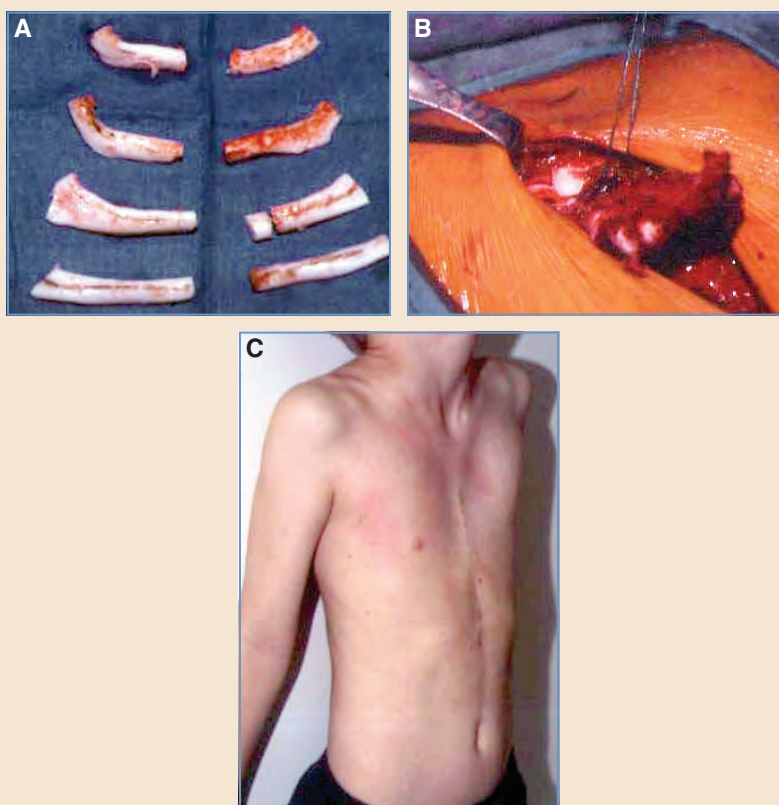


Fig. 58-2 Surgical repair of pectus excavatum. **A**, A subperichondrial costocartilaginous excision. **B**, Circumferential suture fixation with retrosternal cartilage graft incorporated into the posterior osteotomy site. **C**, The patient is shown postoperatively.

modifications include the placement of a permanent vascularized rib strut behind the sternum for support,⁶⁴ the use of a bioabsorbable strut made from poly-L-lactide,⁶⁵ resection of the lower four or five ribs bilaterally and placement of a steel strut,⁶⁶ the insertion of rigid plates for sternal fixation,⁴⁶ a sternal turnover with stabilization and contour,⁶⁷⁻⁷⁰ rib resection with a presterneal suspension support bar,⁷¹ and maintenance of the sternal position with transcutaneous wires connected to an external brace.⁷²

Endoscopic Sternal Repair and the Nuss Procedure

Endoscopic repair is on the horizon.⁷³ A thoracoscope and surgical instruments are inserted through small, 3 mm stab wounds to transect the rib cartilages, to place a retrosternal strut, and to perform an anterior wedge osteotomy. The Nuss procedure corrects sternal angulation without resecting costal cartilages. Nuss et al⁵⁹ described this procedure in a 10-year review published in 1998. It involves inserting a convex steel bar inserted through small, bilateral thoracic incisions and then under the sternum. Modifications to this procedure have been described, including a subxyphoid approach with central fixation to the costal margins.⁷⁴ Multiple comparisons of surgical techniques, primarily between the modified Ravitch procedure and the Nuss procedure, have been made.⁷⁵⁻⁷⁷ They demonstrate no clear difference in outcomes, including success rates, major complications, and patient satisfaction.

The Nuss Procedure

In a Nuss procedure, a thoracic epidural catheter is placed preoperatively for postoperative pain relief. Whenever possible, this is done with the patient awake before the induction of anesthesia to optimize placement. The patient is brought to the operating room, and general endotracheal anesthesia is induced. Prophylactic antibiotics that provide good coverage of skin flora are given. The patient is positioned symmetrically on the operating room table, with shoulders abducted to 90 degrees and elbows bent upward to 90 degrees. This allows bilateral access to the lateral chest wall. A sheet or blanket is rolled and placed longitudinally behind the center of the back so that the shoulders fall backward slightly. The neck, chest, and upper abdomen are prepared with a sterile cleaning solution; the preparation continues down to the operating room table on both sides of the chest. The entire prepared area should be accessible after the patient is draped.

The deepest part of the deformity is marked, and marks are also made in line with this point on either side of the sternum—bilaterally at the midclavicular lines and at the midaxillary lines. The line described by these marks must be square with the patient's chest. If the deepest point of the sternum is inferior to the xiphoid, the lower end of the sternum is marked.

A pectus bar template is chosen to develop the mold for the pectus bar. (All instrumentation for this procedure is available from W. Lorenz Surgical.) The ends of the chosen aluminum template, once shaped, will extend from one midaxillary line to another. The template is molded to the patient's chest wall laterally, leaving the central portion nearly straight. The corresponding stainless steel pectus bar is bent to the same shape using a bar-bending tool. The very ends (2.5 cm) of the bar should not be bent. With the ends straight, the side-stabilizing crosspieces can be properly seated.

Transverse incisions measuring 2.5 cm each are made in the lateral chest wall bilaterally, centered on the midaxillary lines in the plane previously marked. Superior and inferior skin flaps on the serratus muscle fascia are developed, allowing plenty of room for the stabilizing crosspieces. Subcutaneous tunnels on the muscle fascia are then developed from the lateral incisions to the highest ridges of the anterior chest, usually at about the midclavicular lines. A 30-degree thoracoscope is introduced into the left chest, and the chest is gently insufflated with CO₂ (to 5 cm H₂O) to help collapse the lung. This scope is used to view the thoracic organs while the tunneling device is passed behind the sternum.

A tunneling device, or introducer, is passed from the patient's right side through the subcutaneous tunnel to the anterior chest. The introducer enters the chest just medial to the highest ridge through the selected intercostal space. As the introducer enters the chest, it is rotated 180 degrees to hug the back of the sternum as it is passed behind the sternum toward the left chest. This is done while observing through the thoracoscope, and it should be performed very carefully, because this is the most dangerous part of the procedure. The surgeon must stop and try another path if any resistance is met as the tunneler is advanced behind the sternum. Once the tunneler is safely in the left chest, it is again passed anteriorly just medial to the highest ridge into the left-sided subcutaneous tunnel through the appropriate intercostal space. The tip of the introducer is advanced through the subcutaneous tunnel out to the left lateral chest wall incision. Three sizes of introducer are available, depending on the size of the patient.

A double length of umbilical tape is threaded through the eye on the end of the introducer. The introducer is withdrawn back through both sides of the chest and out through the right lateral incision, bringing the tapes with it. The properly curved pectus bar is securely attached to one of the tapes, leaving the second tape as a backup, should the first tape break during passage of the bar. Using traction on the tape from the patient's right side, while observing through the thoracoscope, the pectus bar is advanced upside down through the pathway created. Once the bar has passed through the tunnels and is exiting both lateral incisions, it is turned over using the bar-turning tool. This pushes the sternum and anterior chest wall into the desired position.

Once the bar is properly positioned, the chest wall is corrected. If the correction is deemed to be adequate, the stabilizing pieces can be placed. However, if the correction is not ideal, it may be necessary to place a second bar. This most commonly occurs in older patients who have a lengthy deformity involving both the upper and lower portions of the sternum. If needed, a second bar is typically placed two intercostal spaces above or below the first bar.

Once the correction is adequate, lateral stabilizing crosspieces are placed on both sides of the pectus bar. Although the manufacturer states that only one stabilizer may be needed per bar, bar rotation has been a frequently reported complication; therefore we recommend using two stabilizers per bar. The bar must be derotated to place the stabilizers, but the bar is then rotated once again into the desired position. The bar should sit in the channels in the stabilizers, with the flat side of the stabilizer facing the chest wall. The stabilizers are secured to the chest wall so that they reside at least 1 to 2 cm from the end of the bar. The stabilizers are secured to the pectus bar and to the chest wall using two No. 4 stainless steel wires on either side, medial and lateral to the stabilizer. The stabilizers are then secured to the chest wall using 1-0 Prolene sutures above and below each side of the bar. Large Valsalva breaths are given to evacuate the pneumothorax.

The wounds are inspected, and hemostasis is meticulously confirmed. The subcutaneous tissue of the lateral incision is securely closed with absorbable suture. The wounds are closed with subcuticular absorbable suture and occlusive dressings. A chest radiograph is obtained in the operating room to assess for pneumothorax and to document the alignment of the bar.

Postoperative Care

Postoperatively, pain is controlled with a combination of epidural and intravenous analgesia. Older children and those with a more rigid chest wall have more postoperative pain. Epidural analgesia for 3 to 5 days postoperatively reduces the need for narcotics and the attendant risks associated with their use. Incentive spirometry is actively encouraged, and careful early ambulation is initiated. Patients are weaned from epidural analgesia when tolerated and transitioned to oral pain medications. They are discharged once they are ambulating and eating and pain is well controlled with oral analgesics. A postoperative bowel program is almost always necessary.

Activity is limited for the first 6 weeks after surgery. Good posture is encouraged, and we frequently employ a figure-of-eight clavicle fracture brace to encourage good posture. This is worn night and day for the first 6 weeks. Generally, the pectus bar remains in place for 3 years.

After 3 years, the patient is returned to the operating room. With the patient under general anesthesia, the entire chest is again prepared and draped, allowing access to both lateral chest wall incisions. The previous incisions are opened, and the ends of the pectus bar and the attached stabilizers are exposed and dissected free. The permanent sutures and previously placed wires are removed. A thick scar and even some bony formation around the stabilizers are often present. The stabilizers are removed from the bar. Using the bar-removal and bar-bending tools, the ends of the bar are carefully straightened enough to allow the bar to slide out of one or another of the incisions. Once the bar is removed, a local anesthetic is injected, and the soft tissues and skin are closed in layers with absorbable sutures. This procedure can frequently be performed on an outpatient basis.

Treatment Outcomes and Complications

Haller et al⁵⁶ reported a 95% success rate in 664 patients and 5% mild to moderate recurrence. Kelly et al⁵⁵ reported a similar success rate in 1215 patients, with a 1.4% recurrence rate. Sacco Casamasima et al⁷⁸ reported similar success results in 336 patients, with modifications to the Nuss procedure reducing recurrence rates to 1.7%. Long-term results after a Nuss procedure have remained excellent over 10 years (96.5%), with removal of the support bar in most patients, and an incidence of recurrence of 3%.⁵⁹ The outcome for pectus excavatum repair (see the accompanying video) is

generally excellent when the cartilages are properly resected and the sternum elevated and fixated. According to one study,⁷⁹ complications included wound infection, fluid collection, fracture, migration of hardware, costal cartilage protrusion, pneumothorax, and bar displacement, which was the most common complication. The mortality rate was very low (0.5%).

PECTUS CARINATUM

Pectus carinatum is an anterior convexity of the chest wall and sternum. It is the second most common anterior chest wall deformity after pectus excavatum. Some consider it the antithesis of pectus excavatum. The pathogenesis of both deformities is attributed primarily to the overgrowth of the costal cartilages, with both anomalies described as different manifestations of the same process.^{80,81} If the overgrown cartilages push the sternum inward, pectus excavatum develops, whereas, if the sternum is displaced forward, the result is pectus carinatum.

History and Epidemiology

Pectus carinatum accounts for 6% to 22% of all deformities of the chest wall; the incidence of a familial deformity in these patients is 26% to 33%. The prevalence is estimated as 0.6% to 0.7%.^{82,83} Unlike pectus excavatum, these deformities are usually not noticed in childhood and become apparent during rapid pubertal growth. The male/female ratio is 4:1.⁸⁴ Three categories of morphologic presentation of pectus carinatum are described, based on the location of the maximal convexity along the sternum: (1) keel chest (chondrogladiolar prominence), (2) pouter pigeon breast (chondromanubrial prominence), and (3) lateral pectus carinatum. Keel chest is characterized by sternal elongation and inferior sternal xiphoid prominence. It is commonly associated with bilateral depression of the lower costal cartilages. Pouter pigeon breast has a more superior manubrial sternal prominence with a reduction of the normal angle of Louis, creating a depression of the lower two thirds of the sternal body. Lateral pectus carinatum has a unilateral overgrowth and protrusion with rotation of the sternum to the contralateral side. Associated abnormalities include scoliosis, Marfan syndrome, Poland syndrome, Morquio disease, Noonan syndrome, LEOPARD syndrome, and neurofibromatosis.

History and Assessment

Similar to patients with pectus excavatum, patients with pectus carinatum present for cosmetic correction of their chest wall deformity, because many have a distorted body image.^{85,86} The workup is similar for both. Bronchopulmonary changes have been described in patients with pectus carinatum.^{87,88} Much like pectus excavatum, no concrete evidence shows an improvement in cardiopulmonary status after surgical correction, but subjective improvement has been reported.⁷²

Nonsurgical and Surgical Treatment Options

Nonsurgical options for patients with pectus carinatum seeking cosmetic correction consists of bracing. Haje and Bowen⁸⁹ first described the use of bracing for these patients in 1976 and published long-term results in 1992. Patients usually wear a brace for most of the day for 4 to 8 months, and then transition to a more relaxed schedule. Recent literature suggests that this form of treatment is increasing in popularity.⁹⁰ Recently, Martinez-Ferro et al⁹¹ reported a high success rate among compliant patients.

Multiple surgical approaches have been described for correction of the pectus carinatum deformity. Recently, Schaarschmidt et al⁹² described a minimal-access hybrid technique using endoscopic assistance. Del Frari and Schwabegger⁹³ described a muscle-splitting technique involving the placement of bioabsorbable plates and postoperative bracing. Fonkalsrud⁹⁴ reported a progressive decrease in costal cartilage resection over his 37-year experience, with maintenance of consistent cosmetic results. Kálmán⁹⁵ described a correction procedure analogous to the Nuss procedure for excavatum repair that required neither a cartilage resection or a sternotomy, although long-term results have not been evaluated.

Correction of keel chest pectus carinatum consists of a bilateral resection of the abnormal costal cartilages along the entire extent of the deformity, a transverse sternal osteotomy at the upper end of the deformity, and fracture of the posterior lamina, leading to depression of the sternum. Surgical access is similar to that of open Ravitch-type excavatum repairs, although commonly less extensive. The xiphoid process is left in situ undisturbed and scored posteriorly or partially removed. The sternal construct is secured by suture fixation to the rectus abdominis muscle. The pectoralis muscles are advanced and reunited presternally. A light compression dressing further secures the corrected sternal position.

The steps to correct a pouter pigeon breast are similar to those described for repairing pectus excavatum. The deformed costal cartilages are resected bilaterally, the prominence of the manubriosternal junction is flattened using an osteotome, a transverse V-shaped sternotomy is made at the vertex of the angle of Louis (with a second linear osteotomy at the level of the deepest sternal depression), and the sternum is bent appropriately and detached from its perichondrial and mediastinal attachments. Mesh can be placed behind the sternum for support. The sternum is then covered with pectoralis muscles sutured together presternally. The xiphoid process is reattached to the sternum.

The correction of lateral pectus carinatum depends on the extent of cartilaginous overgrowth. If only a few unilateral cartilages are involved, an incision can be made over them and the protuberance removed subperichondrially. The resection should be more radical than conservative. If cartilage overgrowth is extensive, a short parasternal segment of the cartilages on the contralateral side should also be resected.

Treatment Outcomes and Complications

Surgical treatment outcomes are excellent regardless of the method used. Complications are similar to those for pectus excavatum repair and are uncommon. Recurrence is associated with conservative resection. Growth and development of the chest wall are usually normal after the repair.

JEUNE SYNDROME

The rare thoracic deformity known as *Jeune syndrome* is characterized by skeletal dysplasia, including a narrow immobile thorax and micromelia, with respiratory and renal manifestations. Respiratory manifestations vary widely from respiratory failure and death in infancy to a latent phenotype without respiratory symptoms.

History and Epidemiology

In 1955 Jeune et al⁹⁶ reported two siblings with severely narrowed thoraxes. They explained their condition as familial asphyxiating thoracic dystrophy. The incidence of Jeune syndrome is estimated at 1 in 100,000 to 1 in 130,000 live births. Most patients die in infancy and early childhood; very few mature to adulthood.

Genetic and Etiologic Factors

The genetic pattern of Jeune syndrome is autosomal recessive; 45% of cases are reported in siblings. Virtually all cases are diagnosed by 6 months of age.⁹⁷ Genetic counseling is indicated. The parents of a child affected with Jeune syndrome are obligatory carriers and have a 25% chance of having a second afflicted child. The bony abnormalities are thought to result from in utero disturbance of endochondral bone formation.

Functional Implications

The narrow thorax is motionless, and respiration is entirely abdominal. Considerable supraclavicular and suprasternal retraction of the intercostal space may be present on inspiration. The anatomy includes a long, narrow, and abnormally small thorax with reduced thoracic cage capacity and hypoplastic lungs.⁹⁸ The thorax is narrow transversely and sagittally. The fourth through ninth ribs bend inward, usually at the costochondral junction. The small thorax usually improves with age for those who survive early childhood. The limbs have variable micromelia and short digits with bulbous terminal phalanges and occasional postaxial polydactyly of the hands and feet.

Other associated anomalies include retinal degeneration, intestinal malabsorption, renal failure, hypertension, liver abnormalities, heart disease, and pancreatic abnormalities.⁹⁹

Nonsurgical and Surgical Treatment Options

Supportive medical care may involve mechanical ventilation of the lungs, which is urgently implemented in the most severe cases. In less severe cases, it may be required after several bouts of pneumonia with subsequent ventilatory failure. Phenotypically, this latent group of patients has a chest wall configuration identical to that of neonates with early, severe problems.¹⁰⁰ However, their respiratory compromise is mild and rarely requires surgical correction. Surgery is indicated only in the most severe cases, in whom failure to intervene will result in progressive pulmonary damage and death.

Chest reconstruction requires preoperative planning with CT scans and plain chest radiographs. Pulmonary function and renal function should be evaluated before general anesthesia is induced.¹⁰¹ Thoracic reconstruction is performed by a longitudinal sternotomy to increase intrathoracic volume and fixation with bone grafts, a methylmethacrylate prosthesis, steel struts, or rib grafts.^{102,103} In one technique, horizontal K-wires are placed across the sternal defect, coated with methylmethacrylate, and linked vertically with additional acrylic¹⁰⁴ (Fig. 58-3). A methylmethacrylate prosthesis can be made before surgery, thus saving considerable anesthesia time. The prosthesis supplies support along the entire length of the sternal edges to prevent herniation of the heart and lungs. It may be replaced with a larger prosthesis as the child grows.

Davis et al¹⁰⁵ described lateral thoracic expansion for correction of asphyxiating thoracic dystrophy in patients with Jeune syndrome. This procedure is performed without splitting the sternum. The chest wall is enlarged by dividing the ribs and underlying tissue in a staggered fashion to ensure that either rib or periosteum covers the lung. Titanium plates maintain the rib separation. Waldhausen et al¹⁰⁶ have expanded the vertical expandable prosthetic titanium rib (VEPTR) procedure for application in Jeune patients. Vertical titanium struts are inserted,

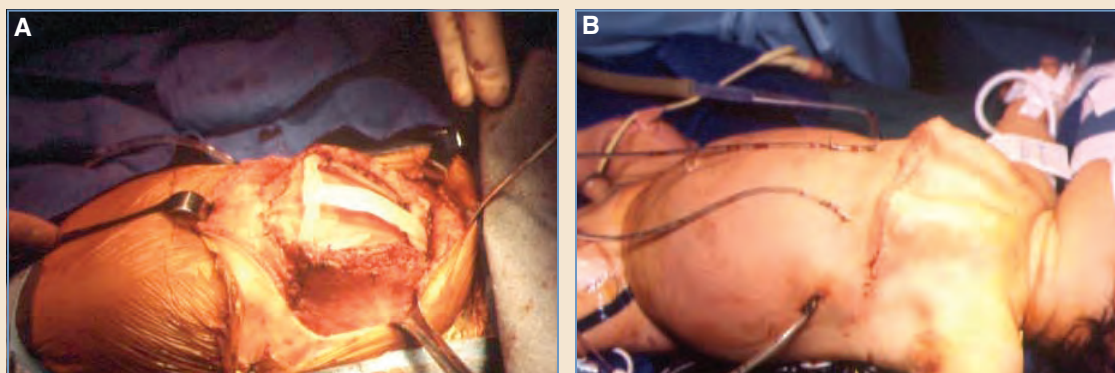


Fig. 58-3 A, An intraoperative view of a patient with Jeune syndrome undergoing placement of an acrylic-reinforced methylmethacrylate K-wire strut. B, An immediate postoperative view of a Jeune syndrome repair.

attached to the ribs and transverse processes of the spine, and progressively expanded to increase the intrathoracic volume. Although the vertical expandable titanium rib procedure has been successful in patients with pediatric thoracic insufficiency from other causes, its use in Jeune syndrome patients has led to controversial improvements in pulmonary function, and long-term results of this approach are not yet available.¹⁰⁶

Kikuchi et al¹⁰⁷ modified the Nuss technique, described previously, for use in older patients with more mild forms of Jeune syndrome. Muthialu et al¹⁰⁸ reported using a single-stage bilateral treatment involving titanium plating for rapid expansion, although long-term results have not been evaluated. Conroy et al¹⁰⁹ published a case of sternal distraction for severe neonatal Jeune syndrome, which was reported to be successful at 8 months.

In addition to surgical correction of the chest deformity, dialysis and, often, renal transplantation are indicated for renal failure.

PENTALOGY OF CANTRELL

Cantrell syndrome,¹¹⁰ also known as *pentalogy of Cantrell*, consists of the following anomalies:

- An anterior diaphragmatic hernia
- An upper omphalocele
- Ectopia cordis
- Intracardiac anomalies
- A bifid sternum

Cantrell syndrome often presents as an incomplete constellation, with hallmark anomalies of an omphalocele and ectopia cordis. Various other anomalies have been described in association with Cantrell syndrome, including a cleft lip/palate, hydrocephalus, encephalocele, limb defects, gallbladder anomalies, and polysplenia.¹¹¹ Skeletal and dermatologic manifestations have been reported.¹¹²

History and Epidemiology

Cantrell et al¹¹⁰ first described this condition in 1958 at Johns Hopkins Hospital. It is a rare syndrome; the prevalence is estimated at 5.5 in 1 million live births.¹¹³ The survival rate is less than 40%, and early surgical intervention does not necessarily improve outcome.^{114,115}

Genetic and Etiologic Factors

The cause of pentalogy of Cantrell is not known. Cantrell et al¹¹⁰ originally postulated a developmental failure of lateral mesoderm at about 14 to 18 days of gestation (before or immediately after the differentiation of the primitive mesoderm into the splanchnic and somatic layers), with failure of development of the transverse septum of the diaphragm and failure of migration of the paired mesodermal folds of the upper abdomen ventromedially. Carmi and Boughman¹¹³ hypothesized that pentalogy of Cantrell is the most severe expression of a variety of anomalies involving the development of the ventral midline, accounting for the variable and often incomplete phenotypic presentations.

Functional Implications

Cardiac anomalies include ventricular septal defect, atrial septal defect, tetralogy of Fallot, left ventricular diverticulum, anomalous pulmonary venous return, and patent ductus arteriosus.^{112,113} Sternal malformations are very diverse, including a bifid sternum, absence of the xiphoid process, absence of the lower two thirds, and incomplete formation of the lower half of the sternum.¹¹²

Nonsurgical and Surgical Treatment Options

Pentalogy of Cantrell can often be diagnosed with prenatal ultrasonography within the first trimester of pregnancy, when an omphalocele can frequently be identified. The rest of the pentalogy should be ruled out concurrently and includes the use of prenatal ECG.^{116,117} MRI has been suggested for prenatal and postnatal evaluation to detect more subtle fetal anomalies.¹¹⁸⁻¹²⁰

The surgical treatment of pentalogy of Cantrell is individualized as a result of the variability of defects, and timing depends on a patient's specific defects and the urgency of intervention because of pending respiratory or heart failure.^{115,121} Incomplete and complete pentalogy has been treated both in a single stage and in a staged approach.^{115,122-124} The omphalocele is typically closed as soon as possible. Intracardiac defects are corrected in accordance with the specific defect and its indications. The order of closure of abdominal wall defects and intracardiac defects depends on the size of the abdominal defect and the stability of the cardiac defect; large abdominal wall defects may need to be closed before cardiac surgery, whereas smaller abdominal wall defects can be repaired afterward. However, cardiac lesions that are sufficiently complex may need to be corrected first.¹¹² Sternal clefts are often closed in the neonatal period with sternal banding and autologous tissue (when sufficient); alloplastic mesh or plating can be placed until sufficient autologous material is available.^{125,126} The diaphragm is often plicated, and the heart is returned to the thoracic cavity by fixation of the pericardium to the ventral side of the diaphragm.¹²⁷

KEY POINTS

- Patients with Poland syndrome may have thoracic cage, sternal, muscle, and breast deformities requiring bony reconstruction, the creation of a symmetrical soft tissue, and reconstruction of an aesthetic breast.
- Pectus excavatum is, at a minimum, a cosmetic deformity that can be corrected surgically with an implant and soft tissue augmentation or with bony manipulation using an open, endoscopic, or minimally invasive technique.
- Pectus carinatum can involve the lower anterior chest (keel chest), the upper anterior chest (pigeon chest), or the lateral chest (lateral pectus carinatum). Surgical correction requires bony resection, stabilization, and soft tissue recontouring.

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59

Congenital Defects of the Abdominal Wall

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ongenital defects of the abdominal wall comprise a large spectrum of deformities, ranging from small umbilical hernias to giant omphaloceles and eventration associated with gastroschisis. Also included is a group of patients with varying degrees of congenital abdominal wall laxity, such as those with prune belly syndrome and megacystic microcolon intestinal hypoperistalsis syndrome. These conditions have a wide variety of causes, associated morbidities, and management.

ANATOMY

An umbilical hernia is a central abdominal wall defect involving the linea alba, with persistent herniation of intraabdominal contents into the umbilical stalk (Fig. 59-1). The skin overlying the hernia is intact.¹

An omphalocele, or exomphalos, is a midline defect of the ventral abdominal wall, resulting in extrusion of intraabdominal contents covered by a membrane comprising peritoneum, Wharton jelly, and amnion. The umbilical cord arises from the membranous sac, and the defect may be located in the central, upper, or lower abdomen. Depending on the size and location of the defect, the hernia may contain bowel, liver, and other organs (Fig. 59-2). Omphaloceles are categorized as minor or major (giant). The definition of a giant omphalocele varies between authors and ranges from a 4 to 5 cm defect to the presence of the liver.²

Gastroschisis is characterized by a full-thickness fissure of the abdominal wall, usually to the right of midline and not involving the umbilicus. Frequently, an intact strip of skin intervenes between the umbilicus and the gastroschisis opening. This results in evisceration of intraabdominal contents without a sac or covering. Small intestine is always present, and stomach, colon, and

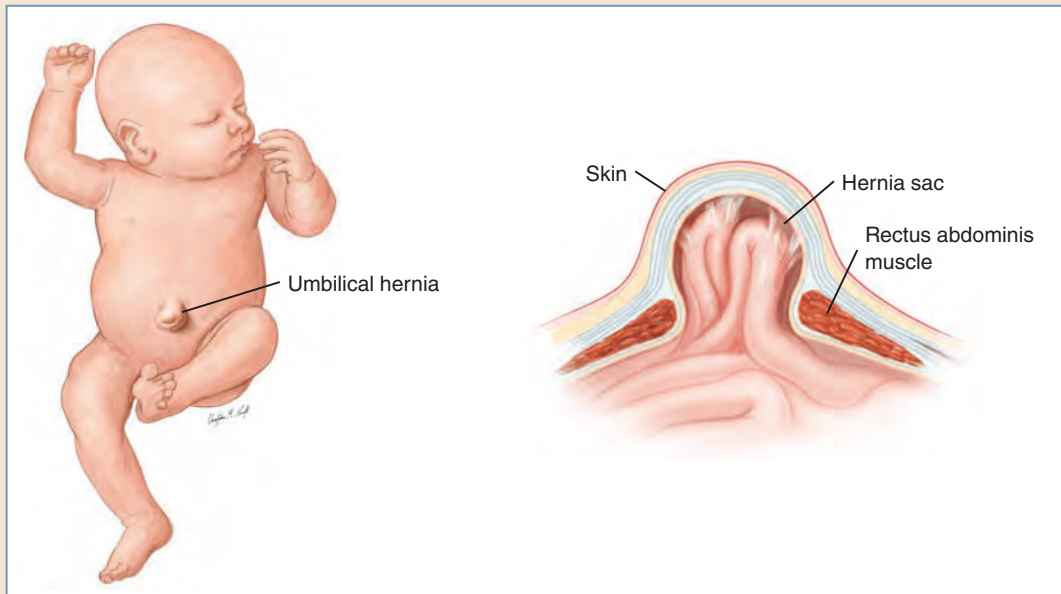


Fig. 59-1 An umbilical hernia is a herniation of abdominal contents into the umbilical stalk, with coverage by intact overlying skin.

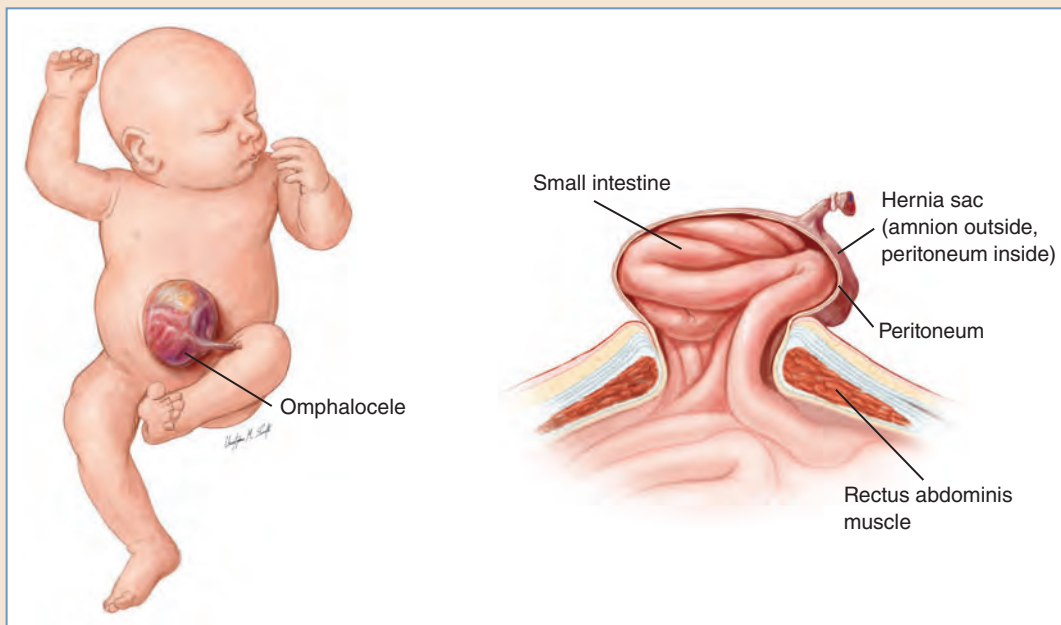


Fig. 59-2 An omphalocele is a midline defect of the abdominal wall, with extrusion of intraabdominal contents that are covered by a thin membrane consisting of peritoneum, Wharton jelly, and amnion.

gonads may be involved. The exposed organs, especially the intestine, have varying severity of serositis and may be covered with a rind thought to be a result of chronic exposure to amniotic fluid (Fig. 59-3). The inflamed bowel frequently causes dysmotility and malabsorption. Gastroschisis is classified as simple or complex, based on the presence of additional bowel anomalies such as atresia, stenosis, volvulus, and perforation.³

Prune belly syndrome (Eagle-Barrett syndrome) consists of deficient musculature of the abdominal wall, cryptorchidism, and urinary tract anomalies (Fig. 59-4).

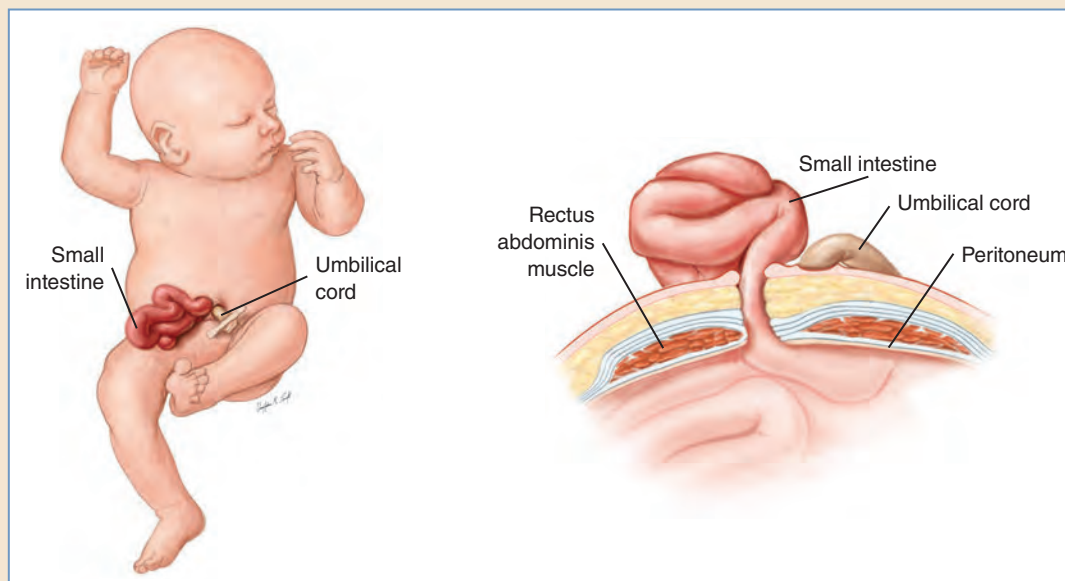


Fig. 59-3 Gastroschisis is a full-thickness fissure of the abdominal wall, with an intact strip of skin between the umbilicus and the gastroschisis opening. Abdominal contents eviscerate and have no sac or covering.

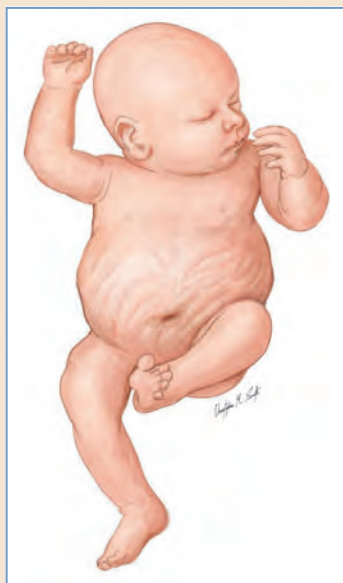


Fig. 59-4 Prune belly (Eagle-Barrett) syndrome consists of deficient abdominal wall musculature, cryptorchidism, and urinary tract anomalies.

Affected neonates can have associated pulmonary dysfunction, constipation, and renal failure. The extent of abdominal wall involvement ranges from mild hypoplasia to complete absence of the abdominal wall musculature.

EMBRYOLOGY AND PATHOGENESIS

During weeks 3 and 4 of gestation, the trilaminar disk of the developing embryo begins to fold in both the craniocaudal (longitudinal) and the transverse directions, forming a more cylindrical structure and isolating it from the embryonic membranes (Fig. 59-5).

At the completion of this folding, the cranial and caudal folds join with both lateral folds to form the ventral body wall. The central umbilical stalk is the only connection to the embryonic membranes. The resulting pleuroperitoneal canals are then divided by the septum transversum into the pleural and peritoneal cavities. In weeks 6 to 12, the rapidly elongating midgut transiently herniates into the umbilical cord as its rate of growth exceeds that of the peritoneal cavity. The

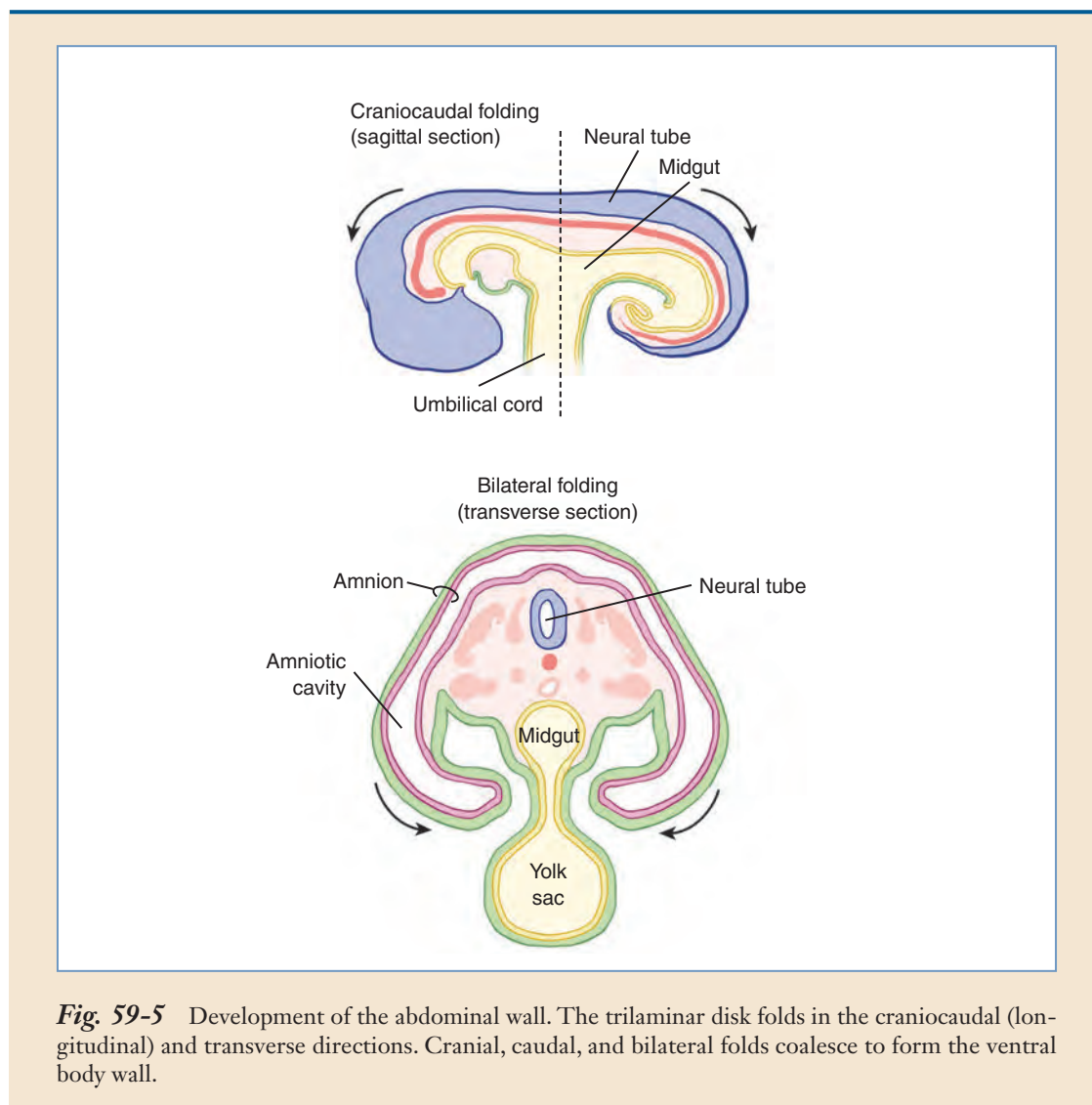


Fig. 59-5 Development of the abdominal wall. The trilaminar disk folds in the craniocaudal (longitudinal) and transverse directions. Cranial, caudal, and bilateral folds coalesce to form the ventral body wall.

midgut undergoes a 270-degree counterclockwise rotation around the axis of the superior mesenteric artery as it returns to the peritoneal cavity.

The development of an umbilical hernia is thought to result from failure of the umbilical ring to close completely. The cause of omphaloceles, however, is thought to be failure of fusion of one or more of the folds. In the most common form, failed fusion of the lateral folds results in a central abdominal defect. Involvement of the cranial fold can lead to an epigastric/thoracic defect known as the *pentalogy of Cantrell*, which includes an omphalocele, a diaphragmatic hernia, a sternal cleft, ectopia cordis, and an intracardiac defect such as a ventricular septal defect (Fig. 59-6) (see Chapter 58).

Similarly, involvement of the caudal fold can lead to a hypogastric/pelvic defect involving the urogenital system—a collection of findings known as the *OEIS complex* (omphalocele, extrophy of the bladder, imperforate anus, and spinal defects)⁴ (Fig. 59-7).

The cause of gastroschisis is not well understood, but it is thought to be a separate entity from omphaloceles. Several theories have been proposed, including thrombosis of the right omphalomesenteric vein leading to abdominal wall necrosis, a failure of physiologic herniation of the rapidly expanding midgut leading to rupture of the abdominal wall, and/or failure of mesodermal formation or penetration.



Fig. 59-6 A stillborn with an omphalocele, a sternal cleft, and ectopia cordis.

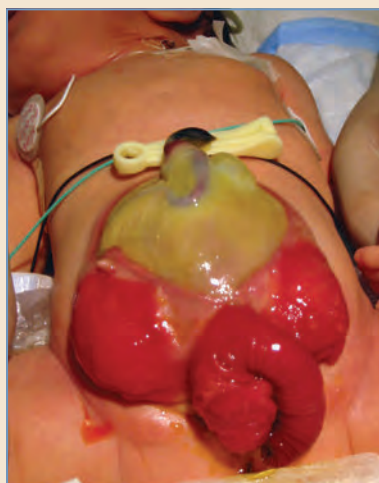


Fig. 59-7 A patient with the OEIS complex consisting of an omphalocele and extrophy of the bladder.

The cause of prune belly syndrome is not well defined but is thought to involve the development of an in utero urethra or bladder outflow obstruction, with resultant pressure effects on the abdominal wall and genitourinary tract. Aberrant mesenchymal development at 6 to 10 weeks of gestation has been suggested as an alternate mechanism.

The timing of the intrauterine event appears to be important, with earlier problems leading to more severe manifestations. The development of an omphalocele is thought to occur at about week 7 to 8 of gestation, with gastroschisis arising at about week 6 to 7. Earlier events at weeks 4 to 6 can lead to failure of fusion in all three planes. The resultant condition, known as a *body stalk defect* or *limb-body wall complex*, involves abnormalities of the head and neck, spine, extremities, and anterior body wall, with direct fusion of organs to the placenta. It carries an extremely poor prognosis.⁵

EPIDEMIOLOGY

Umbilical hernias are one of the most common congenital malformations, with a reported incidence of up to 25%.⁶ Many of these resolve without intervention, and the true incidence may be much higher. The incidence is significantly higher in black infants and in premature infants. Up to 75% of infants weighing less than 1500 g are affected.

The incidence of omphalocele is reported as 1.5 to 3 per 10,000 births and appears to be stable.⁷ Most omphaloceles arise sporadically, although rare familial cases have been reported. Omphalocele has a high incidence of associated anomalies (50% to 70%). Cardiac defects are found in 30% to 50%, whereas chromosomal abnormalities occur in up to 30% of cases. Trisomy 13, trisomy 18, and trisomy 21 are frequently observed. A variety of syndromes are associated with omphaloceles, the most common of which is Beckwith-Wiedemann syndrome.⁸

Gastroschisis is seen in 2 to 3 per 10,000 births, and unlike omphalocele, the global incidence appears to be increasing.⁸ No defined genetic causes have been identified, although the reported risk is 6% if one sibling is affected. It is associated with low maternal age, low BMI, socioeconomic status, prenatal alcohol consumption, smoking, and/or recreational drug use.⁷ In contrast to omphaloceles, associated anomalies are much less commonly seen with gastroschisis, and an abdominal wall defect is the only abnormality in more than 90% of patients⁸ (Table 59-1).

Prune belly syndrome is seen in 1 in 29,000 to 1 in 40,000 live male births. Males are predominantly affected; approximately 3% of all cases occur in females. The incidence of prune belly syndrome in twins (12.2 per 100,000 live births) is significantly greater than that seen in single

Table 59-1 Characteristics of Gastroschisis and Omphalocele

	Gastroschisis	Omphalocele
Defect	Open	Membrane-covered
Defect size	2-5 cm	2-15 cm
Umbilical cord	Left of defect	Center of membrane
Bowel	Inflamed	Normal
Alimentation	Delayed	Normal
Associated anomalies	Uncommon	Common
Prognostic factors	Condition of bowel	Associated anomalies

births. Associated abnormalities include Potter facies, chest and abdominal wall deformities, cardiac murmurs, myocutaneous deformities, and a distended bladder.⁹

PRENATAL DIAGNOSIS AND WORKUP

A prenatal diagnosis of abdominal wall defects allows optimal management of the pregnancy, focused screening for associated anomalies, and early introduction of the parents to the surgical team. Families can be counseled about the condition, associated prognosis, and expected management. After a major abdominal wall defect is diagnosed, surgeons should consider transferring the patient to a hospital with experience and expertise in the care of these complex patients, should none be available at the existing facility.

The mainstay of prenatal diagnosis involves an ultrasound evaluation at weeks 16 to 22 of gestation (Fig. 59-8) and testing for maternal serum alpha-fetoprotein. In combination, these are 80% sensitive for the detection of abdominal wall defects.¹⁰ Maternal serum alpha-fetoprotein is more reliably elevated in gastroschisis compared with omphalocele.¹¹ A distended bladder, distended ureters, large kidneys, extremity abnormalities, cryptorchidism, and oligohydramnios detected on a fetal ultrasound examination may indicate prune belly syndrome.¹²

Fetuses with gastroschisis and omphalocele are at risk for intrauterine growth retardation, premature delivery, and in utero death. Those with an omphalocele should be evaluated for associated structural anomalies, including a diagnostic ultrasound screening for cardiac and renal defects and an amniocentesis for chromosomal abnormalities.

After the diagnosis is made, patients are followed with serial ultrasound examinations to evaluate growth, oligohydramnios, and bowel dilatation. Examinations are performed more frequently in the third trimester. A biophysical profile and fetal nonstress monitoring are indicated for physiologic evaluation.

In general, pregnancies are allowed to go to term unless changes in a fetus' condition dictate earlier delivery, although the timing of delivery is a controversial topic. Vaginal delivery is feasible for most patients with gastroschisis, whereas a cesarean section is frequently performed for patients with giant omphalocele to prevent rupture of the sac.¹³

For prune belly syndrome, in utero percutaneous vesicoamniotic shunting has been described as a means of decompressing the bladder, improving oligohydramnios, and minimizing some of the associated abnormalities.¹⁴



Fig. 59-8 A three-dimensional ultrasonographic image of the abdomen of a baby showing gastroschisis.

MANAGEMENT

With uncomplicated umbilical hernias, incarceration is rare, and small defects (less than 1 cm) generally close spontaneously by the age of 1 to 2 years. Most surgeons therefore recommend a period of initial observation, reserving surgical intervention for persistent or larger defects. The timing of surgery is controversial, although most surgeons agree that it should not be performed before a patient is 2 to 3 years of age.

The initial management of neonates with abdominal wall defects begins with the principles of airway, breathing, and circulation. They are at risk for premature delivery, and the associated bronchopulmonary dysplasia may require intubation and mechanical ventilation. Neonates with a large omphalocele may have associated pulmonary hypoplasia, which further increases the likelihood of respiratory distress. Vascular access should be obtained for fluid resuscitation and intravenous antibiotics, and the umbilical vessels are cannulated if necessary. Ultimately, all babies with gastroschisis require robust venous access for total parenteral nutrition (TPN) supplementation.¹⁵

Gastroschisis

For neonates with gastroschisis or a ruptured omphalocele, the next priority is to minimize evaporative and heat losses from the exposed viscera. Most authors recommend placing the bowel in a transparent plastic bag or wrap,¹⁶ which minimizes insensible losses, helps to control body temperature, and provides some protection while allowing continuous visual monitoring of the exposed bowel. The baby is placed right-side down with the viscera supported, to prevent kinking of the bowel's mesentery. Electrolytes and blood glucose levels are checked and corrected as required. The high fluid losses possible with exposed viscera may require much higher intravenous fluid rates for resuscitation. A nasogastric tube is placed and maintained to suction for decompression.

The bowel is examined for other pathology, including atresia, necrosis, or perforation, and the baby is examined for other associated anomalies. Tight defects of the abdominal wall may require urgent enlargement, whereas volvulus may necessitate urgent reduction. In the absence of these findings, the decision needs to be made whether to attempt immediate reduction and repair or to perform staged (silo) closure.¹⁷ A primary repair is preferred whenever feasible; however, a number of key variables can potentially prevent this option:

- The size of the defect
- The degree of abdominovisceral disproportion
- The condition of the bowel
- Extreme prematurity
- Severe pulmonary disease
- Associated conditions or anomalies

The risk of primary reduction and closure is related to the inevitable increase in intraabdominal pressure resulting from returning the viscera to the abdomen, which in turn may lead to respiratory compromise, renal insufficiency, and abdominal compartment syndrome.¹⁸ As the degree of abdominovisceral disproportion increases, these risks also rise accordingly. Similarly, in extremely premature infants or infants with severe pulmonary disease, the pulmonary status may be quite tenuous, and pulmonary function may be compromised more easily and/or more severely. In these cases, it is often more prudent to undertake staged closure. Several studies have compared the outcomes of primary closure versus silo closure for gastroschisis, looking at overall complication rates, time on the ventilator, time to full feeds, and length of hospital stay. Although the results of these studies can be difficult to interpret because of inherent selection bias, with judicious patient selection the outcomes can be equivalent.^{19,20}

Primary Reduction and Repair

Primary reduction and repair is generally performed with the patient under a general anesthesia with a neuromuscular blockade. A urinary catheter is placed not only to measure urine output but also to periodically measure bladder pressure intraoperatively and postoperatively. Bladder pressure and peak airway pressures are proxies for determining intraabdominal pressure, and significant elevation of this value after reduction of the viscera may indicate that primary fascial closure is not advisable at the first stage.

Bladder pressures greater than 10 mm Hg are considered elevated in children,²¹ and some authors advocate a hard cutoff value of 20 mm Hg in deciding whether to proceed with primary repair. Although parameters have been documented for bladder pressure indicating intraabdominal hypertension and acute abdominal compartment syndrome, there are no absolute values for peak airway pressures that vary significantly between patients, dependent on age and preexisting pulmonary comorbidity. Therefore relative changes in peak airway pressures are used as a proxy for increased intraabdominal pressure and the ability to extubate the patient after surgery. Trending the changes in bladder pressure and airway pressures should be continued into the immediate postoperative period.²²

Near-infrared spectrometry (NIRS) is an additional noninvasive methodology that reliably reflects visceral tissue oxygenation and perfusion in neonates weighing less than 10 kg. Although frequently used on larger babies, its reliability and usefulness in larger babies are controversial.²³

The bowel should be carefully examined before reduction and areas of atresia, stenosis, and vascular compromise noted. Vascular compromise can result from a tight fascial defect, in which case urgent enlargement of the defect is indicated.²⁴ Attempted separation of inflamed, matted loops of bowel before reduction is not advisable, because it frequently results in enterotomy. In the absence of intestinal compromise, the bowel mass is reduced through the defect. The bladder, airway pressures, and NIRS values (if available) should be noted before and after attempted reduction; if pressures are not excessively elevated and NIRS measurements did not decrease, fascial closure can be performed. The skin and subcutaneous fat are elevated circumferentially from the fascia, exposing the fascial defect, which is often quite small. Primary suture repair of the myofascial defect can then be carried out, followed by layered closure of the overlying tissues.

If intestinal atresia is identified, it may be possible to resect the affected segment and perform a primary anastomosis at the same operation. This is dependent on the overall condition of the bowel and the location and extent of the atretic segment. Other options include the formation of an end ostomy for distal atresia or simple reduction of the unrepaired bowel with repair of the abdominal wall, deferring repair of the intestine to a second stage.²⁵ Similarly, areas of bowel necrosis may be treated with resection and primary anastomosis under ideal conditions or may require ostomy formation or tube decompression.²⁶

After closure, the abdomen should remain soft; parameters reflecting intraabdominal pressure and visceral perfusion mentioned previously should be stable, and acidosis should not develop. If any one of these criteria is not met, then primary repair is abandoned and a staged approach chosen instead.²⁷ Some authors have advocated that, to minimize physiologic embarrassment, viscera may be reduced at the bedside with mild sedation and spontaneous ventilation,²⁸ whereas others have recommended a staged closure with silo placement in all cases.²⁹

Staged Silo Repair

Schuster¹⁷ first described the creation of a silo, consisting of Teflon sheets sewn together to cover the exposed bowel and sutured to the abdominal wall under moderate tension. This was covered with mobilized skin flaps, which were periodically reopened to facilitate serial tightening and removal of the Teflon. Allen and Wrenn²⁷ subsequently modified the technique to use Silastic sheets instead of Teflon, leaving the construct exposed to facilitate monitoring of the viscera and

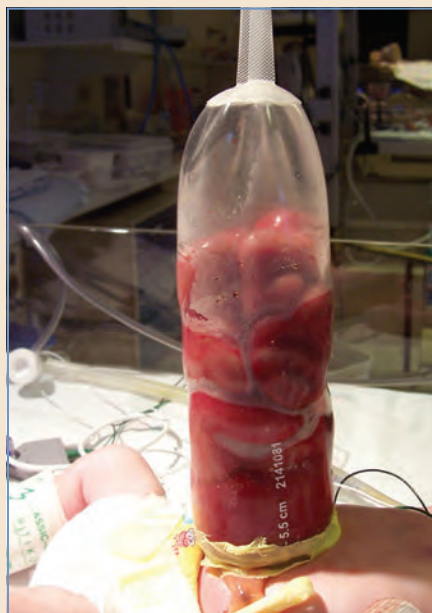


Fig. 59-9 A prefabricated Silastic silo is suspended above the patient to allow gradual reduction of the viscera into the abdominal cavity.

serial tightening. This became the standard technique for staged repair until the introduction of a prefabricated Silastic silo (Dow Corning) coupled to a spring-loaded ring (Bentec) in the 1990s.²⁹ With this device, the spring-loaded ring will hold the silo in place as long as the fascial ring is intact. It can be placed at the bedside without anesthesia and is suspended overhead (Fig. 59-9).

The now-protected bowel becomes less swollen and edematous as the neonate physiologically diureses over the next several days. This, coupled with gravity, allows the bowel to gradually reduce back into the peritoneal cavity. The silo can be serially tightened by placing a clamp across the top to hasten the reduction. This process may take 1 to 14 days, after which the skin, fascia, or both can be closed. Silo placement remains the standard of care for gastroschisis patients in whom primary closure is not feasible. Some authors have reported on a “plastic” sutureless closure technique, in which the viscera are reduced as before, and the umbilical cord is trimmed and laid over the gastroschisis defect and covered with an adhesive dressing.³⁰ Proponents of the technique cite improved scarring and umbilical positioning; however, resultant ventral hernia rates are high (up to 84%), and this has not been universally accepted.

Omphalocele

Evaporative and heat losses are less with omphalocele compared with gastroschisis, because of the presence of the sac covering the viscera. Surgery often is less urgent, and managing associated anomalies may take precedence over definitive correction of the omphalocele. Initial management, as for gastroschisis, involves airway stabilization and temporary coverage of the defect. The omphalocele sac can be covered with moist gauze and a plastic wrap, and the neonate should undergo a complete evaluation for associated anomalies, including undiagnosed genetic syndromes. An echocardiogram and renal ultrasound are carried out.³¹

After this initial management, attention is turned to the omphalocele. As for gastroschisis, the decision to attempt early reduction and fascial closure is guided by the size of the defect and the degree of abdominovisceral disproportion. The overall condition of the infant and comorbidities, particularly pulmonary hypoplasia, are also important factors.

For small omphaloceles smaller than 2 to 4 cm, early reduction with primary fascial closure is the benchmark and has been shown to result in less sepsis, fewer operations, and lower mortality.²⁸ The amniotic sac is removed and the bowel examined for associated atresia and malrotation. After reduction, the operation proceeds much the same as for gastroschisis, with elevation of skin flaps, freeing of the fascial edges, and primary myofascial suture repair. Intraabdominal and airway pressures guide the surgeon both intraoperatively and postoperatively.²²

For large omphaloceles and cases in which the intraabdominal pressures preclude early reduction, staged closure may be undertaken. A Silastic silo or Teflon pouch can be used, as for gastroschisis. However, the umbilical ring tends to be less robust in an omphalocele; therefore the spring-loaded ring is ineffectual, and the device is sutured directly to the fascia. The sac is left intact throughout this process as a barrier to infection and subsequent adhesions, and serial tightening can be performed. After the viscera are returned to the abdomen, the infant is taken to the operating room for removal of the device and repair of the fascial defect. Mesh repair of the fascia has been described³²; however, we prefer not to use this whenever possible. Intraoperative tissue expansion, as originally described by Sasaki,³³ can be used to obtain moderate gains in abdominal domain, facilitating closure of larger defects.

The umbilical cord is invariably ligated during an omphalocele repair, leaving no umbilicus. A pseudumbilicus may be created with a purse-string or local skin flap technique, either at this operation or a later stage.

For giant omphalocele defects and for infants too unstable to undergo extensive surgery, a delayed repair is performed. In this technique, the omphalocele defect is covered with skin, but no attempt is made to reduce the abdominal contents or close the fascia. Gross³⁴ originally described the mobilization of bipedicle flank skin flaps to cover the omphalocele sac, with skin grafting to the donor sites. At the time, this represented a major advance in the surgical management of these patients. However, in common with all delayed repairs, it did not address the potentially massive ventral hernia that invariably resulted.

Delayed repair initially can be managed nonoperatively, and in many centers this has become the standard for treating giant omphaloceles. The omphalocele sac is treated with a topical agent that promotes eschar formation and is allowed to reepithelialize from the edges, a process that may take up to 10 weeks. Initial reports used mercurochrome, alcohol, or silver nitrate; each was effective but associated with potential toxicity.³⁵ Silver sulfadiazine (Silvadene) has emerged as an excellent alternative with less potential toxicity.³⁶ Dressings are performed twice daily until epithelialization is complete and may potentially be completed on an outpatient basis in selected patients.³⁷ Gentle compressive wraps can be placed as an adjunct during and after reepithelialization to support the abdominal mass and to encourage gradual return of the abdominal contents into the abdomen in preparation for a definitive hernia repair, usually 6 to 12 months later.³⁸ Alternatively, skin coverage may be expedited using skin grafts directly on the omphalocele sac, preventing the need for a prolonged period of dressing changes.

The management of omphaloceles requires a team approach, and multiple surgical specialties may be involved. In particular, low omphaloceles associated with cloacal extrophy or OEIS complex will require a pediatric surgeon, plastic surgeon, urologic surgeon, and an orthopedic surgeon to work closely together. Similarly, high omphaloceles associated with sternal, thoracic, or cardiac anomalies will require the input of a pediatric and a cardiothoracic surgeon. A complete discussion of the management of these complex patients is presented in Chapter 58.

Prune Belly Syndrome

Similarly, patients diagnosed prenatally with, and those who are suspected of having, prune belly syndrome should be transferred to a hospital with pediatric specialty services, including an experienced obstetric and neonatal team, pediatric urologists, and reconstructive surgeons. The extent of abdominal wall weakness in prune belly syndrome ranges from mild hypoplasia to complete absence of the musculature. Milder manifestations of this condition may benefit from fascial plication, whereas moderate to severe deficiencies require more extensive surgical intervention. A variety of approaches have been described, most of which involve excision of redundant fascia and placement of mesh. Lesavoy et al³⁹ reported on a series of 20 patients with moderate to severe prune belly syndrome, who underwent an overlapping “double-breasted” fascial repair with concurrent urologic repair. The authors reported good results, with only three patients having a reoperation for right-sided fascial laxity within the long follow-up period (mean 20 years).

In the most severe forms of prune belly syndrome, the abdominal musculature is completely absent. The outcomes of abdominal reconstruction in this patient group are uniformly poor, and surgeons should consider an abdominal wall transplant for patients who are candidates for a kidney transplantation or a posterior rectus sheath–liver vascular composite allotransplantation if a multivisceral transplant, including the liver, is indicated.^{40,41}

POSTOPERATIVE CARE

The initial postoperative management of infants with abdominal wall defects varies by condition, severity, and the presence of associated anomalies or comorbidities. In general, mechanical ventilation is continued for a few days postoperatively. This is particularly true after closure of larger omphaloceles, where it is beneficial in the short-term to maintain neuromuscular blockade (paralysis) and sedation during the early postoperative period. During this time, nasogastric tube decompression is continued. Continued monitoring of bladder and airway pressures is often warranted, and judicious fluid administration may help to limit edema formation.

Once bowel edema has subsided and intraabdominal pressures have decreased, then paralysis and sedation can be safely lifted and the patient weaned from the ventilator as tolerated according to comorbidities, particularly pulmonary hypoplasia. The same holds true after repair of gastroschisis; however, the loss of abdominal domain is usually less than with giant omphaloceles; therefore resultant intraabdominal pressures may be less extreme.

Infants with gastroschisis uniformly require central venous access and total parental nutrition, because the exposed intestine invariably demonstrates dysmotility and absorptive dysfunction. Enteral feeding is deferred until 2 to 3 weeks after repair in many cases. The presence of bowel atresia or other types of complex gastroschisis may delay this even further.³

Omphalocele patients may require TPN in the short term, depending on the extent of the omphalocele and associated morbidity. However, the intestinal dysmotility and absorption abnormalities seen with gastroschisis are generally not present in omphalocele patients; therefore the need for TPN and the duration of treatment are generally less.

OUTCOMES

The long-term outcomes for infants born with congenital abdominal wall defects have improved dramatically since the 1960s, in large part because of the introduction of TPN,¹⁵ advances in ICU care, and the recognition that staged repair may be necessary in larger defects.¹⁷ In the absence of associated anomalies, the prognosis after an abdominal wall repair is generally excellent.

As discussed previously, infants with gastroschisis can have delayed bowel function and prolonged TPN requirements, and a period of catch-up growth is the rule after successful feeding has begun. With prolonged TPN comes the possibility of associated complications, including line sepsis and TPN-associated cholestasis. Survival from gastroschisis is greater than 90% and generally related to the degree of intestinal loss.⁴² The length of hospital stay is also related to bowel dysfunction, with tolerance of enteral feeds delayed longer than 2 months in up to 25%.⁴³ In addition, necrotizing enterocolitis can develop after repair of gastroschisis, contributing to delayed intestinal function and subsequent malabsorption or short-gut syndrome.⁴⁴ In the long term, most patients are able to tolerate full enteral feeds and catch up in growth satisfactorily.⁴⁵ However, prolonged TPN in infants may be further complicated by oral aversion to food, requiring long-term behavioral and swallowing therapy while nutrition is maintained through a nasogastric or gastrostomy tube. It has been reported that the method of closure does not affect survival, length of stay, or duration of TPN, whereas duration of mechanical ventilation appears to be slightly shorter with use of a silo versus primary closure.¹⁹

The overall survival for neonates with an omphalocele is significantly lower, at 60%, and is more highly dependent on the presence of associated anomalies. Survivors tend to have higher rates of chronic morbidity, including pulmonary hypoplasia, gastroesophageal reflux, and failure to thrive.⁴⁶

Despite these issues, the quality of life in the survivors of both gastroschisis and omphalocele have been reported as good to very good and not significantly different from that of the general population.⁴⁷ As an example, a significant proportion of these patients report the appearance of the umbilicus, or lack thereof, as one of their most prominent long-term concerns.^{46,48}

The mortality of patients with prune belly syndrome depends on the degree of pulmonary hypoplasia and the development of renal failure. In the past, approximately half of the patients with prune belly syndrome died in the neonatal period. The advent of in utero vesicoamniotic shunts has significantly improved early survival.¹⁴ Although many of these patients had improved renal and bladder function, approximately a third of these infants ultimately required dialysis and/or renal transplantation.

ALTERNATIVE TREATMENTS AND MANAGEMENT OF SECONDARY HERNIA DEFECTS

The treatments presented previously are the standard of care for patients with congenital abdominal wall defects from gastroschisis or omphalocele. However, several additional strategies have been reported with varying degrees of success.

The use of negative pressure wound therapy (wound vacuum-assisted closure [VAC]) has been described as an alternative to silo treatment, facilitating gradual reduction of the extracoelomic contents in small case series.⁴⁹ The reported benefits include continual pressure on the extracoelomic mass, preoperative reduction of the viscera, and splinting of the omphalocele contents. The authors also described its later use over an intact skin graft, reportedly increasing abdominal domain in preparation for definitive ventral hernia repair in a patient 8 months of age. Although these results, and those of other small series, are promising, negative pressure wound therapy has not been extensively studied in these patients and has not been universally accepted.

The inevitable loss of intraabdominal domain seen with giant omphaloceles has also been addressed through the use of tissue expanders. These devices potentially allow a rapid increase in abdominal domain, creating space for reduction of the abdominal contents without a dangerous increase in intraabdominal pressure. Some have described placement of the expanders in the subcutaneous plane, between the internal oblique and the transversus abdominis muscles, and intraperitoneally. Complications, including infection, deflation, extrusion, and displacement or malposition, have limited tissue expanders' efficacy and routine use.^{50,51}

Component separation has also been described for managing patients with omphaloceles or gastroschisis, with promising initial results. In a recent case series, Levy et al⁵² reported on the use of component separation in nine children with a median age of 1.1 years at operation. In four patients, the technique was used for recurrent ventral hernias. Component separation was used as a first-line treatment to primarily close the fascia in three neonates, with no evidence of recurrence within a follow-up of 16 months. The authors used acellular dermal matrix (ADM) reinforcement in eight of nine patients. Although component separation successfully closes these large ventral abdominal wall defects, the long-term effects of disuse atrophy of the external oblique muscles that frequently occurs with component separation have not been evaluated.

The use of ADM and other bioprosthetic materials such as small intestinal submucosa (Surgisis; Cook Surgical) is growing in popularity in many areas, including abdominal wall reconstruction. Purported advantages of these materials over prosthetic mesh include tissue incorporation and revascularization, the ability to grow with the child, and the ability to use it in a contaminated field.⁵³ In a report on three patients with complex gastroschisis, small intestinal submucosal bioprosthetic Surgisis was used to patch the fascial defect and was covered with a negative pressure dressing until granulation and skin graft or reepithelialization.⁵⁴ Similarly, Beres et al⁵⁵ described the use of Surgisis for fascial repair in nine patients with omphaloceles and two patients with gastroschisis. They reported a 38% recurrent hernia rate. In an earlier report, Kapfer and Keshen⁵⁶ reported on the use of Alloderm (LifeCell) human ADM bioprosthetic for early fascial reconstruction in three patients with a giant omphalocele. The Alloderm material was left partially exposed in two patients and dressed with serial cadaveric allograft dressings. The authors reported no infections and no fascial dehiscences.⁵⁶ Although these materials may work well to prevent dehiscence in the short term, when used to bridge a gap, they will invariably cause a bulge or result in recurrent herniation in the long term.

Similarly, an ADM can be used during the intermediate steps of planned, multistage repair in the secondary management of hernia defects resulting from delayed closure of omphalocele defects, as described in the following case.

In patients undergoing solid organ transplant, an abdominal wall transplant represents another potential method of addressing loss of abdominal domain. Levy et al⁵² reported on a series of patients undergoing both a small bowel and abdominal wall transplant, one of whom was a child with gastroschisis. Similarly, Ravindra et al⁴⁰ published their experience of a concurrent transplant of small bowel, pancreas, liver, and posterior rectus sheath in a patient with gastroschisis, short bowel syndrome, loss of abdominal domain, and TPN-induced cholestasis. They performed a multivisceral transplant using the posterior rectus sheath attached to the liver through the falciform ligament. The posterior rectus sheath component of this vascular composite allograft was used to repair the abdominal wall fascial defect.

CASE EXAMPLE

This 23-month-old boy was born with a giant omphalocele and associated pulmonary hypoplasia. Because of pulmonary hypertension and other complicating factors, his omphalocele defect was treated in a delayed fashion with Silvadene but was complicated by bowel perforation. He was managed with placement of a Silastic silo and Vicryl mesh (Fig. 59-10, *A*), and skin grafting was performed several weeks later. He was transferred for further management, presenting with a large ventral hernia under a healed skin graft and severe loss of domain (Fig. 59-10, *B* through *D*).

A staged repair was planned, which included insertion of intraabdominal tissue expanders to increase abdominal cavity capacity and placement of subcutaneous tissue expanders (Fig. 59-10, *E*) to provide healthier skin closure over a temporary ADM that was planned to be inserted at a second stage.

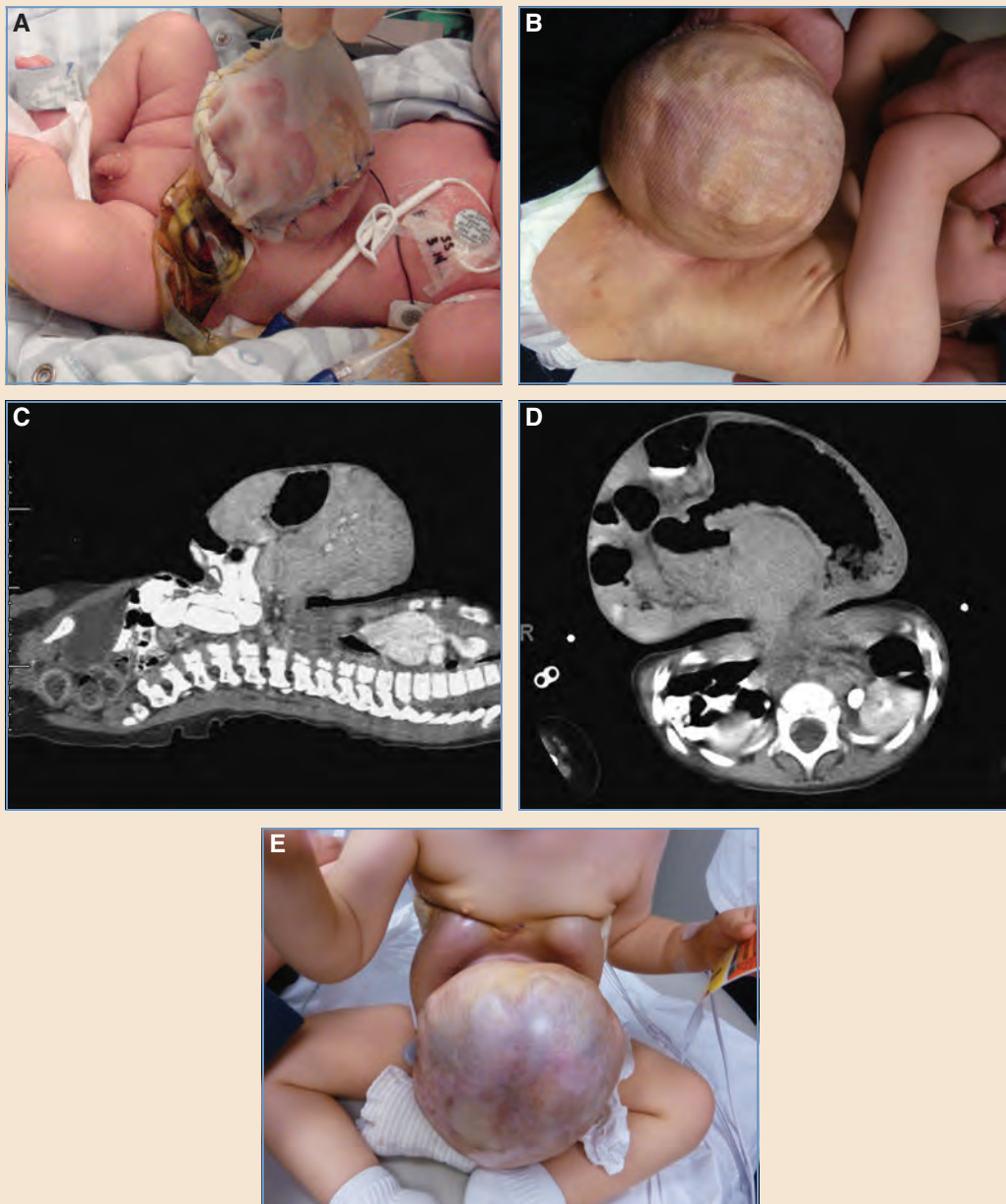


Fig. 59-10 **A**, A silo was sutured in place. **B**, He had a giant omphalocele at 2 years of age, with healed skin graft coverage. **C**, Lateral CT showed the small size of the abdominal cavity versus the large size of the omphalocele. The AP diameter of the abdominal cavity was about 1.5 cm, and the AP diameter of the omphalocele was 9.3 cm. The transverse diameter of the abdomen was 11.8 cm, as opposed to the transverse measurement of the omphalocele of 13 cm. **D**, Coronal CT demonstrated the discrepancy of the abdominal capacity versus the size of omphalocele. **E**, The patient is shown 1 month after expander placement.

Continued

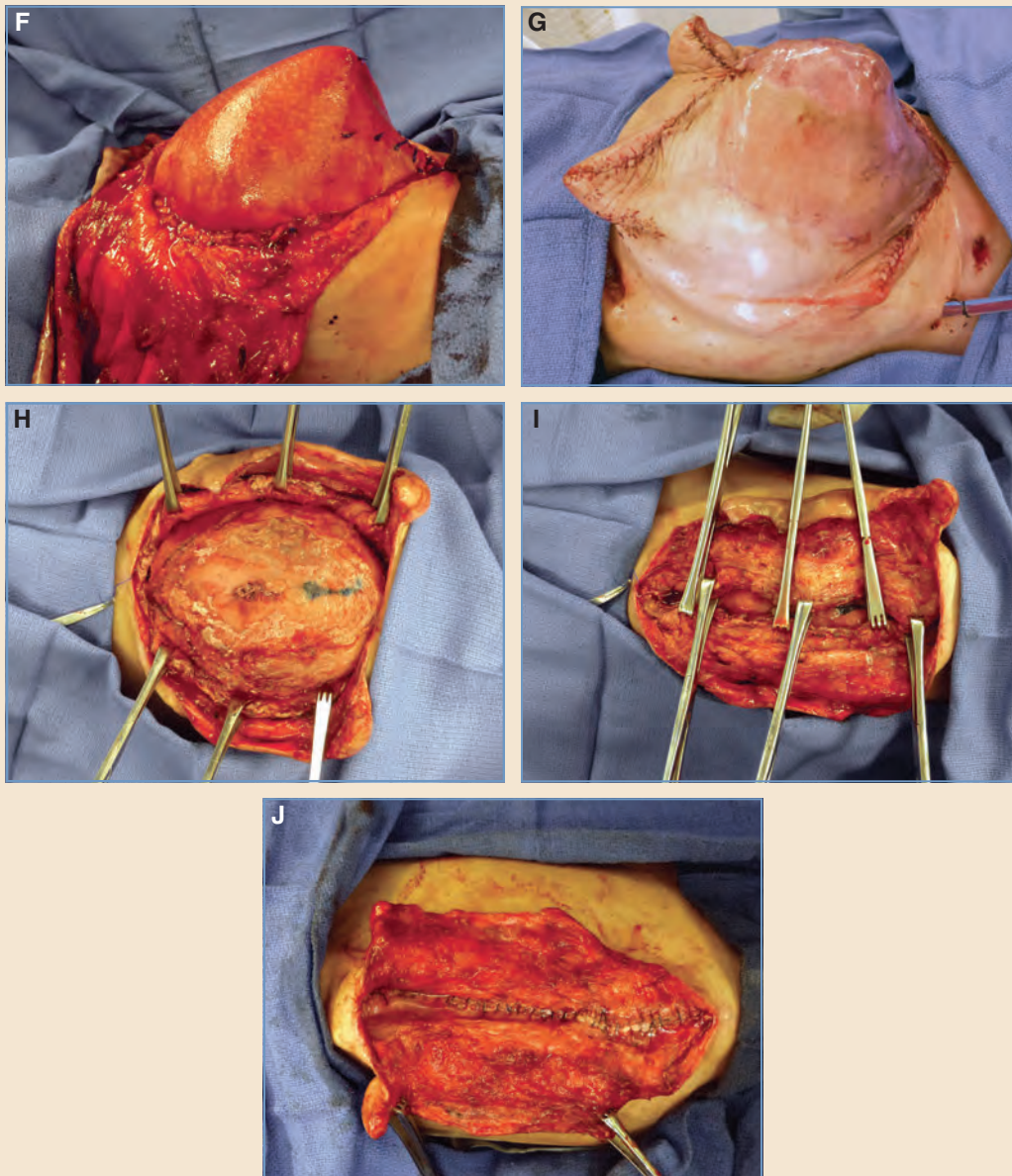


Fig. 59-10, cont'd F, Viscera were partially reduced into the expanded abdominal cavity and covered with an ADM. G, Residual skin graft and the expanded skin were approximated over the ADM. H, Fascial edges are secured with Lahey clamps. I, With intermittent stretch-relaxation, the edges were brought toward midline. J, Fascia to fascia closure.

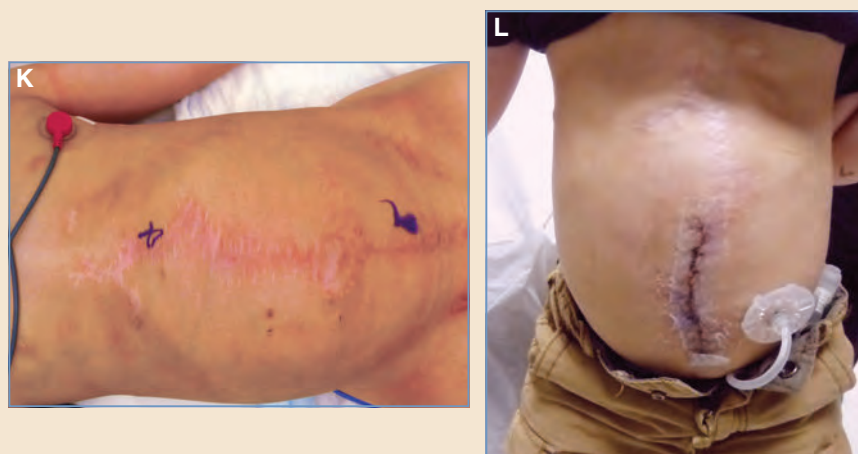


Fig. 59-10, cont'd K, Six months later, the patient was prepared for a gastrostomy tube placement. L, He is shown 1 month after placement of the gastrostomy tube and 18 months after the initial insertion of tissue expanders.

The plan was complicated by extrusion of one of the subcutaneous expanders, necessitating early removal and deflation of one of the intraabdominal expanders, because it slipped into the extraabdominal component of the omphalocele. Despite this, the second stage proceeded with partial reduction of his viscera and placement of a temporary porcine ADM (Fig. 59-10, F).

Because of the loss of one of the expanders, only a part of the skin graft could be excised. Residual skin graft and the expanded skin were approximated over the ADM (Fig. 59-10, G), and the bladder and airway pressures remained stable throughout. With partial reduction of the liver, his pulmonary hypertension improved significantly. One week later, the patient underwent plication of the porcine ADM Strattice mesh (LifeCell) and further excision of the skin graft. A small area of wound breakdown resulted in exposure of the ADM, which was removed 3 weeks later and replaced with a smaller ADM underlay patch. On completion of this repair, increased bladder pressure and oliguria were noted intraoperatively, necessitating release of the reconstruction and the use of a larger ADM patch. The skin was reapproximated over this.

This reconstruction was again complicated by minor wound breakdown, and 3 months later he developed an infection of the abdominal wall. The ADM was removed. The abdominal wall was expanded intraoperatively using a stress-strain-relaxation technique, and primary fascial approximation was achieved (Fig. 59-10, H through J).

Intraoperative monitoring included bladder pressures and the use of a NIRS device placed on the skin over one kidney. He tolerated the procedure very well, and was extubated on the first postoperative day.

Seven months later, at 38 months of age, the patient underwent a repeat laparotomy with gastrostomy tube placement for management of persistent feed aversion (Fig. 59-10, K). He had no evidence of recurrent hernia. On attempted reclosure of the abdomen, the fascia felt thin; therefore a unilateral myofascial advancement flap was performed to reinforce the central closure.

Six months postoperatively, the patient is progressing well with no evidence of herniation, and his oral intake and respiratory status are steadily improving (Fig. 59-10, L).

CONCLUSION

In the broad spectrum of congenital abdominal wall defects, the two most common are omphaloceles and gastroschisis. The outcomes of these conditions have dramatically improved over the last 50 years because of improved technique selection, ICU care, and TPN. For patients with gastroschisis, intestinal function is generally poorer, but overall survival is greater than 90%. For those with omphaloceles, gastrointestinal function is less impaired, but overall morbidity and mortality are much higher because of the high incidence of associated anomalies. Despite associated morbidities, quality of life can be very good in survivors and will continue to improve as surgical management strategies continue to evolve.

KEY POINTS

- Congenital abdominal wall defects comprise omphaloceles, gastroschisis, and prune belly syndrome.
- Serial antenatal ultrasounds are critical after the diagnosis of an abdominal wall defect.
- Percutaneous in utero vesicoamniotic shunting significantly improves early survival and minimizes associated deformities in patients with prune belly syndrome.
- Early fluid and heat losses can be significant with gastroschisis or ruptured omphalocele, and coverage of the exposed bowel is a surgical emergency.
- Evaporative losses are much less with omphalocele than with gastroschisis.
- The incidence of associated anomalies is much higher with omphalocele, and these should be actively sought.
- The ultimate goal of treatment is adequate coverage of the viscera.
- The size of a defect, the degree of abdominovisceral disproportion, and associated morbidities dictate the method of treatment.
- Staged or delayed repair may be necessary in selected cases.
- The physiologic impact of an omphalocele is related to its size and the presence of associated anomalies.
- In gastroschisis, the bowel does not function properly, but the child rarely has other serious issues.
- Alternative treatment methods such as tissue expansion, negative pressure wound therapy, and the use of bioprosthetic mesh can help to optimize outcomes.

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Pediatric Breast Anomalies

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Pediatric breast anomalies span a wide range of conditions. Some, such as polythelia, are relatively common and present as isolated abnormalities, whereas others, such as amastia, are rare and may present with a constellation of congenital issues. Treatment of breast abnormalities involves decisions about the type and timing of treatment and consideration of patients' immediate and future aesthetic requirements.

Pediatric breast anomalies may be classified as hypoplastic, hyperplastic, or deformational.^{1,2} Hypoplastic conditions are characterized by breast tissue paucity and include hypoplasia, tuberos breast, Poland syndrome, athelia, and amastia. These conditions require augmentation procedures. Hyperplastic anomalies are characterized by excess breast tissue and include polythelia, polymastia, gynecomastia, juvenile hypertrophy, and giant fibroadenomas. These conditions require treatment with excision techniques. Deformational anomalies include alterations in breast growth resulting from surgical, traumatic, and burn scars, or chest wall asymmetry resulting from scoliosis or pectus excavatum. These deformities may require a combination of augmentation and excision procedures.

NORMAL BREAST DEVELOPMENT

The breast develops during the sixth week of gestation from an anlage of ectodermal cells along the milk lines, or primitive mammary ridges, which extend from the axilla to the groin. By the tenth week of gestation, the upper and lower parts of these ridges atrophy; at the level of the fourth interspace, the middle or pectoral ridges persist and later develop into breast tissue. The areola develops in the fifth month of gestation, and the nipple appears shortly after birth.³

After birth, circulating maternal estrogens may cause breast tissue enlargement. As maternal estrogens are metabolized, the breast tissue involutes, remaining quiescent throughout childhood. During childhood, the breast consists of epithelial-lined ducts with surrounding connective tissue. At the onset of puberty, hormonal influences in females result in breast tissue growth; that is, estrogens cause ductal and stromal tissue growth, and progesterone causes alveolar budding and lobular growth.⁴ Thelarche, the onset of breast growth, occurs at an average age of 11 years (normal range 8 to 15 years of age) and usually precedes menarche by at least a year. Tanner⁵ described breast development as proceeding through stages I through V, with breast maturity occurring in stage V. Nipple size differentiation occurs between Tanner stages IV and V.⁶ Breast growth may be variable, but is generally complete by 16 to 18 years of age.

CLASSIFICATION OF BREAST ABNORMALITIES

Hypoplastic Breast Abnormalities

Breast hypoplasia (with an intact nipple-areola complex) can be unilateral or bilateral.

Tuberous breast deformities are characterized by breast hypoplasia (Fig. 60-1), and include the following⁷:

- A deficiency in the base diameter
- Breast tissue herniation into the areola
- A deficient skin envelope
- Elevation of the inframammary fold

Several classification systems have been developed to describe tuberous breasts.⁷⁻⁹ Meara et al⁹ proposed a three-tier system in which type I tuberous breasts have medial quadrant hypoplasia, type II have lower medial and lateral quadrant hypoplasia, and type III have severe breast

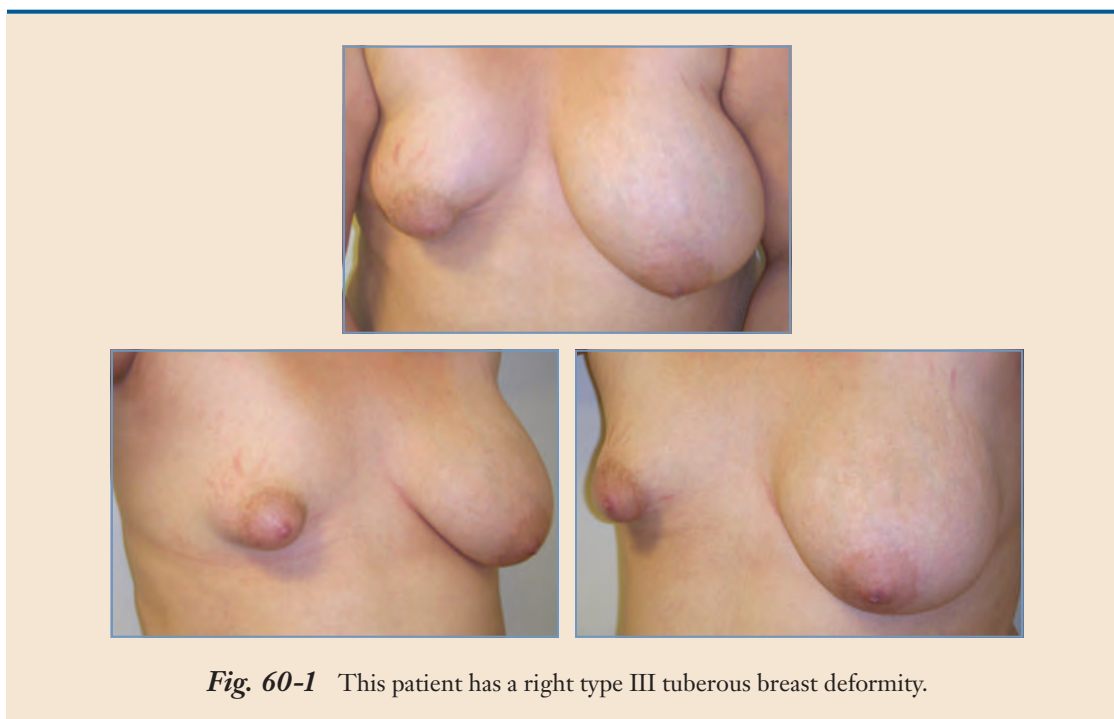


Fig. 60-1 This patient has a right type III tuberous breast deformity.

Table 60-1 Tuberous Breast Grading System

Type	Breast Base	Inframammary Fold	Skin Envelope	Breast Volume	Ptois
I	Minor constriction	Minor elevation medially	Sufficient	Minimally deficient	None
II	Moderate constriction	Elevation medially with minor elevation laterally	Insufficient inferiorly	Mild to moderate deficiency	Minimal to moderate
III	Severe constriction	Elevation of the entire fold	Insufficient circumferentially	Severe deficiency	Severe

**Fig. 60-2** This young male has left Poland syndrome. His pectoralis muscle is absent.

constriction^{9,10} (Table 60-1). As the grade of tuberosty increases, the deformity involves a progressive elevation of the inframammary fold, an increasing paucity of skin, a decreasing breast volume, and increasing ptosis.

Poland syndrome is a chest wall deformity affecting 1 in 20,000 to 1 in 30,000 live births that usually results in a more significant breast hypoplasia than tuberous breast (Fig. 60-2). Poland's initial description of the deformity included pathognomonic absence of the sternal head of the pectoralis major and brachysyndactyly of the ipsilateral hand, which can be variable.¹¹ Other possible associated findings include absent ribs, chest wall depression, athelia or amastia, absent axillary hair, and deficient subcutaneous fat. The anomaly is usually unilateral and sporadic, but rarely bilateral or familial.^{10,12,13} A similar condition, anterior thoracic hypoplasia, consists of a unilateral sunken chest wall with hypoplasia of the breast and a superiorly displaced nipple-areola complex. These individuals have a normal sternal position and a normal pectoralis muscle¹⁴ (see Chapter 58).

Athelia (absence of the nipple), amazia (absence of the mammary gland), and amastia (absence of the nipple and gland) are rare congenital hypoplastic anomalies^{2,15,16} (Fig. 60-3). Three groups of amastia patients have been described: (1) those with bilateral absence of the breast secondary to congenital ectodermal defects, (2) those with unilateral absence of the breast (a variant of



Fig. 60-3 This male infant has athelia.

Poland syndrome), and (3) those with bilateral absence of the breast.¹⁶ Amastia associated with congenital ectodermal defects affects males and females and is characterized by abnormalities of the skin and appendages, including the teeth and nails. Bilateral absence of the breast may occur as an isolated abnormality, or it may be associated with clefts of the palate and upper extremity anomalies. The defect can be sporadic or familial.¹⁶

Hyperplastic Breast Anomalies

Hyperplastic breast anomalies are characterized by excessive breast tissue. The most common form of hyperplasia seen by pediatric plastic surgeons is polythelia, which is the presence of supernumerary nipples or entire nipple-areola complexes. This anomaly results from failure of the superior or inferior primitive mammary lines to fully atrophy. Polythelia occurs in males and females and has a reported incidence as high as 5.6%.^{17,18} It generally occurs sporadically and in rare instances may be associated with nephrourologic anomalies;¹⁹ familial cases have also been reported. Polythelia can occur at any point along the embryonic milk line from the axilla to the groin² (Fig. 60-4).

Polymastia, the presence of more than two breasts, is a rarer condition than polythelia and may occur anywhere along the embryonic milk line.²⁰ It is usually sporadic, but familial cases have been reported; latent cases may become noticeable during puberty, pregnancy, or lactation. Polymastia can occur as an isolated finding or may present with congenital renal anomalies^{2,21} (Fig. 60-5).

Gynecomastia, enlargement of the breast in males, affects up to 65% of males 14 to 15 years of age. It is initially characterized by fibroblastic stromal and ductal proliferation producing a palpable infraareolar breast bud, and it generally resolves spontaneously without surgical intervention.²² Gynecomastia persists in 10% of affected boys at 17 years of age.²³ If the condition persists beyond a year, fibrosis and hyalinization occur with regression of epithelial proliferation, increasing the likelihood that surgical intervention may be required. Simon et al²⁴ proposed a three-grade classification system for gynecomastia in which grade I is characterized by slight breast enlargement without skin redundancy and for which reassurance that the condition should resolve without intervention is appropriate. Grade IIA is characterized by moderate breast enlargement without skin redundancy; grade IIB involves moderate breast enlargement with marked skin redundancy; and grade III has marked breast

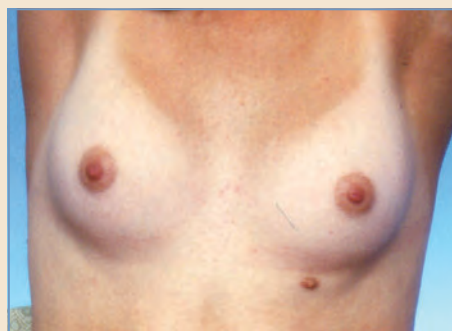


Fig. 60-4 This patient with left polythelia has an accessory nipple just below the inframammary fold.

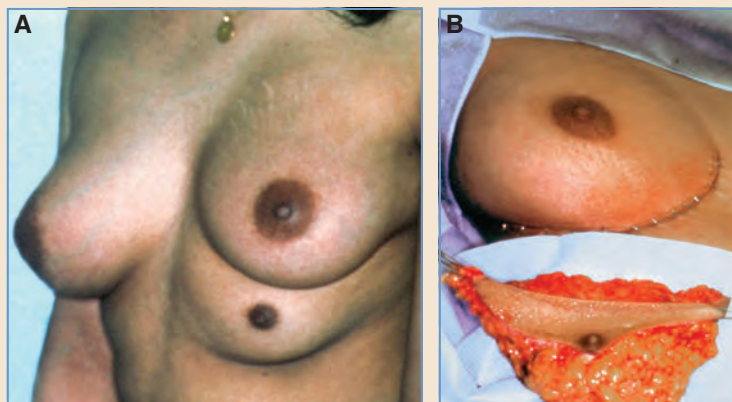


Fig. 60-5 A, Left polymastia. B, An intraoperative view of the resected tissue.

Table 60-2 Gynecomastia Grading System

Grade	Breast Size	Skin Amount
I	Slight breast enlargement	No redundancy
IIA	Moderate breast enlargement	No redundancy
IIB	Moderate breast enlargement	Marked redundancy
III	Marked breast enlargement	Marked redundancy

enlargement and skin redundancy²⁴ (Table 60-2). An endocrine evaluation to rule out a reversible cause of gynecomastia, especially testicular and adrenal tumors, is recommended.

Pediatric breast hypertrophy, or juvenile hypertrophy, has an unknown cause. It is not associated with endocrine abnormalities, and patients otherwise exhibit normal growth. Prepubertal hypertrophy is very rare and generally bilateral, whereas virginal hypertrophy, which develops after puberty, may be either unilateral or bilateral. Patients can have asymmetry from unilateral hyperplasia, bilateral symmetrical hyperplasia, or a combination of hypertrophy and hypoplasia.²⁵



Fig. 60-6 A, This patient had a giant fibroadenoma. B, The excised lesion. C, The postoperative result.

Giant fibroadenoma, a rare form of hypertrophy, is a benign, discrete lesion that presents during puberty as a unilateral, rapidly growing breast mass (Fig. 60-6). These lesions are the result of breast tissue hypersensitivity to normal levels of gonadal hormones. The diagnosis is confirmed with a breast tissue biopsy.

Deformational Breast Anomalies

The most common deformational breast anomalies are growth disturbances caused by scarring, which may be iatrogenic or secondary to trauma and burns. A common iatrogenic injury in females develops from a right anterolateral thoracotomy for cardiac surgery (Fig. 60-7); surgery performed before puberty may result in a mature breast volume difference in 55% of patients and deficiencies in the lower pole of the right breast in 61% of patients, compared with 0% of patients undergoing a median sternotomy for a similar procedure.²⁶ This has led many cardiac surgeons to abandon thoracotomy incisions in female patients.

Severe burns can cause restrictive scarring that affects breast development. Deformities may include loss of the inframammary fold, synmastia (medial confluence of the breasts), upward or downward displacement of the breast mound, and loss of the nipple-areola complex (Fig. 60-8).

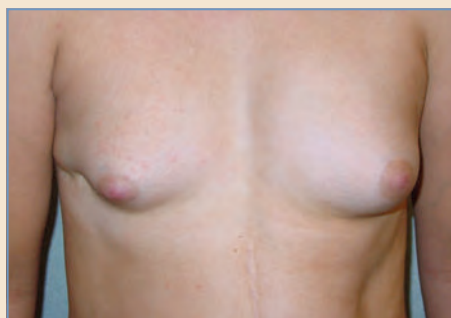


Fig. 60-7 This adolescent girl developed a right breast deformity after an anterolateral thoracotomy.



Fig. 60-8 This patient has a constricted breast deformity resulting from burn scars.

Synmastia is rarely congenital but most often the result of prior surgery or trauma. Spence et al²⁷ first described the condition in 1983. It is characterized by the absence or effacement of the presternal cleft between the breasts.

Deformities of the thoracic cage may result in apparent breast asymmetry or exacerbate actual asymmetry. Patients with pectus excavatum, pectus carinatum, and scoliosis initially can present with breast asymmetry and may require further studies, including CT, to determine whether the spine and/or chest wall deformities also require treatment.

Treatment Strategies

Timing

The timing of reconstruction is important in maturing females. It is often advisable to delay breast surgery until breast development is complete, because breast growth may be variable and asymmetrical, even in healthy individuals.²⁸ Breast growth is presumed to be complete when no change in bra size has occurred for at least a year. Asymmetry of up to a full cup size is considered normal, but more severe asymmetries are pathologic and can result in back, neck, and shoulder pain that improves with breast reduction.²⁹ In patients with asymmetry, operating on the normal breast should be delayed until breast growth is complete. Operating on a growing breast may result in unnecessary surgical revision after growth is completed; however, earlier operation is



Fig. 60-9 A and B, This patient had a bilateral tuberous breast deformity. C and D, She is shown postoperatively after release of breast tissue herniated into the nipple-areola complex and a bilateral augmentation mammoplasty.

indicated and may be beneficial in several situations, such as gross asymmetry of the breast from giant fibroadenomas.

Correct placement of the inframammary fold and nipple-areola complex, balanced and symmetrical breast mounds, and maintenance of normal presternal breast separation are critical for effective reconstruction of any breast deformity. The placement of scars should also be carefully considered to provide the best aesthetic result.

Implants

Breast reconstruction or augmentation is performed with implants, pedicled or free autologous tissue transfer, fat transfer alone, or in combination with other techniques.

Implant reconstruction can be performed as the sole procedure if the volume deficit is small. Stretch marks may result if an overly large implant augmentation is attempted. In patients with skin deficiency, a preliminary procedure to reposition the inframammary fold and insert a tissue expander may be performed to allow expansion of the skin envelope before definitive augmentation.³⁰ When the breast base is severely constricted, as in tuberous breast anomalies, the breast tissue must be incised in a radial fashion to allow the breast envelope to expand to fit over the implant² (Fig. 60-9).

When saline implants are used for reconstruction in severely hypoplastic breasts, especially if submuscular placement is not possible, implant palpability and rippling may result; silicone implants, which are less likely to cause these problems, can give more aesthetically pleasing results. The likelihood for revision surgery, rupture, capsular contraction, and implant interference with mammographic imaging is high. Reconstruction in patients with amastia should be performed cautiously, because aberrant blood supply to the skin can cause unpredictable healing.³¹

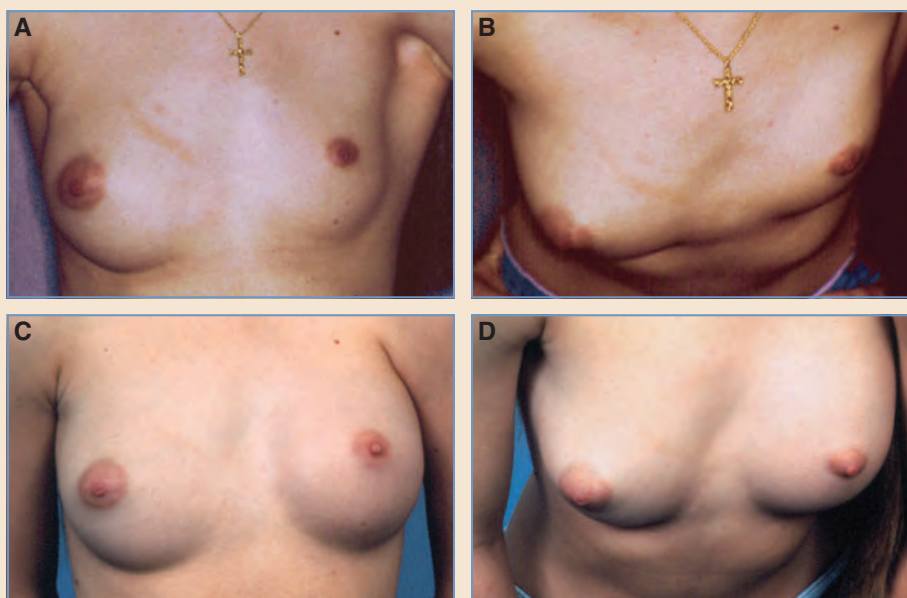


Fig. 60-10 A and B, This patient had Poland syndrome with left breast hypoplasia and absence of the pectoralis major muscle. C and D, After reconstruction with a latissimus dorsi flap and implant placement, she has improved breast symmetry and subclavicular soft tissue contour.

In patients with Poland syndrome, anterior thoracic hypoplasia, and pectus deformities, chest wall deformities can be reconstructed with custom solid silicone implants. This treatment may be most appropriate in male patients with Poland syndrome in whom the use of the latissimus dorsi muscle would be noticeable. Some authors have been critical of these implants, because they are visible under the skin envelope; however, this problem can be prevented by covering the implant with a latissimus dorsi flap³² or with fat grafting.

Tissue Flaps

Some authors recommend the use of inferiorly based flaps to reconstruct breast contour in patients with inferior pole deficiency.³³ This technique results in a smaller breast than can be achieved with implants and is appropriate for milder cases of tuberous breast deformities. Latissimus and rectus abdominis myocutaneous flaps can also be pedicled to reconstruct the absent tissue. A latissimus dorsi flap combined with a breast implant can be a very effective means of reconstructing a severe Poland deformity (Fig. 60-10). Some authors have advocated using endoscopy or even a surgical robot to assist in the harvest and placement of the latissimus dorsi flap^{20,34}; however, we have found that a relatively small incision suffices for this procedure, obviating the need for this technology. Once harvested, the latissimus dorsi flap must be inset carefully along the infraclavicular margin to prevent recurrence of the infraclavicular hollow. We prefer the latissimus dorsi flap because of the high reoperation rates among patients undergoing synthetic chest wall reconstruction without myocutaneous flap coverage.^{1,20}

Free tissue transfer has been useful in patients requiring a large amount of tissue for reconstruction and includes the use of free transverse rectus myocutaneous flaps, deep inferior epigastric perforator flaps, and less commonly used flaps such as a transverse upper gracilis and a

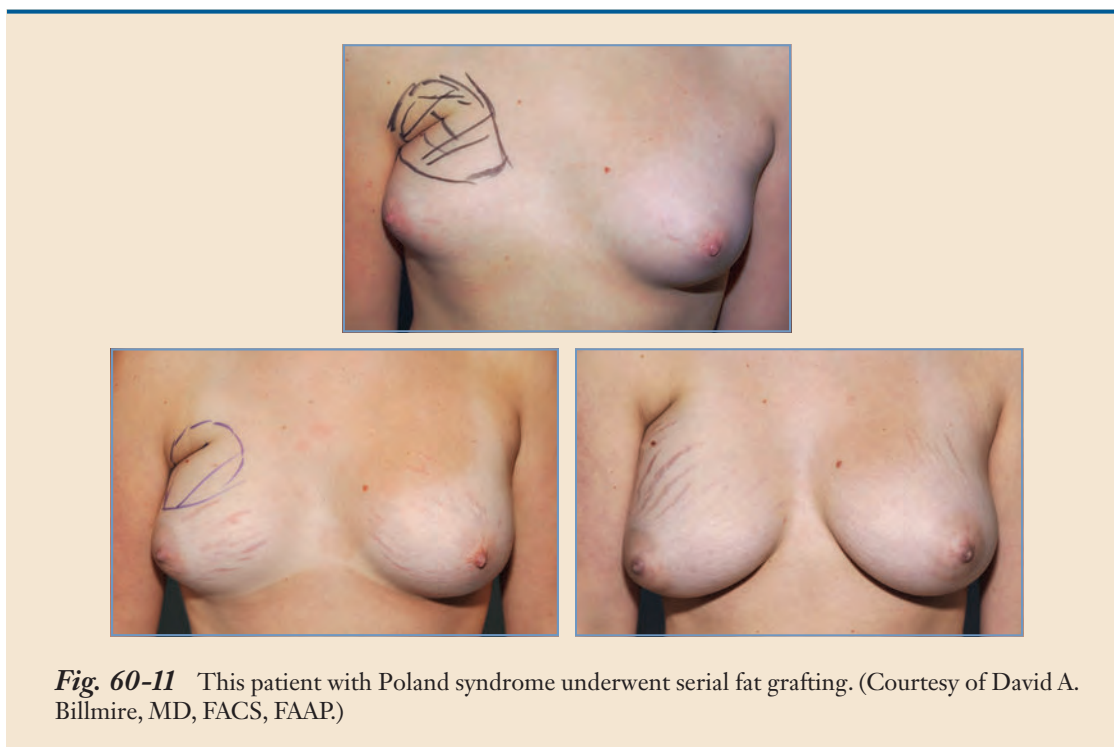


Fig. 60-11 This patient with Poland syndrome underwent serial fat grafting. (Courtesy of David A. Billmire, MD, FACS, FAAP.)

Rubens.³⁵ These are complicated procedures with significant risks; therefore they should be delayed until breast development is complete to prevent unnecessary revisions. Tissue transfer prevents the problem of anomalous blood supply to the chest wall skin and may be especially useful for severe scarring of the breast skin, as in burn scar reconstruction.

Fat Grafting

The technique of micrografting autologous fat is relatively new but has been used in many different applications in plastic surgery over the past decade.^{36,37-39} Fat grafting for reconstructing the breast mound or for correcting subtle irregularities (such as camouflaging the edge of an implant) addresses not only the problem area but also may reduce an overweight donor region such as the abdominal wall. Transferring large volumes of estrogen-producing fat to the growing mammary gland has an unknown effect on oncologic risk. However, large studies of adult cancer patients have failed to demonstrate an increase in breast cancer risk after fat transfer.³⁶

Fat grafting has been used to correct breast hypoplasia and asymmetry and to camouflage underlying chest wall asymmetries.⁴⁰ It also effectively corrects tuberous breast deformities⁴¹ (Fig. 60-11). A benefit of fat grafting is that the procedures can be performed in series, allowing gradual expansion of the skin envelope, which leads to a more natural shape that is not achievable with traditional implant augmentation. Reconstruction for breast asymmetries can begin at a younger age, as soon as patients express an interest in surgery; the lipofilling may be tailored to keep pace with the normally developing breast. This has psychological advantages for patients who might otherwise be encouraged to wait with an uncorrected deformity until breast maturity.

In our practice, the survival rate of fat grafts in pediatric patients has been high compared with rates in adult patients. This allows dramatic correction in two to three operations but may cause problems in patients who subsequently gain weight, and breast reduction may be required in severe cases. In most patients, however, the increase in overall body fat with adolescent growth allows a better correction of the deformity as the fat expands beyond what was surgically transferred.

Excision

Polythelia is best treated with simple excision of the accessory nipple or nipple-areola complex. Typically, when this is performed before puberty, no glandular breast growth occurs in the area, but periareolar hair follicles may be present. If multiple nipple-areola complexes are present on the breast itself, determining which complex is associated with glandular/ductal tissue can be challenging. An MRI may be needed to determine which nipple-areola complex to preserve. Breast reduction techniques can be used to reposition the areola appropriately.

Polymastia is treated with excision, usually with excellent cosmetic results. The most common presentation of polymastia is bilateral axillary breast tissue without duplication of the nipple-areola; this is easily excised with an axillary incision. Follow-up is recommended because of the possibility of developing cancer in retained breast tissue at the excision site. When polymastia is present on the thorax, both breasts are usually relatively hypoplastic, and the inframammary fold may be asymmetrical. Resection of one nipple-areola with preservation of the associated breast tissue allows repositioning of that tissue so that the breast volume can be preserved for improved symmetry.

Giant fibroadenomas that are small can be excised directly; however, larger ones may distort the breast and require breast reduction techniques.⁴² The timing for surgery is based on the onset of the rapid growth phase of the fibroadenoma; surgical excision is often necessary before breast development is complete because of the gross discrepancy in breast size (see Fig. 60-6).

Treatment of Gynecomastia

Gynecomastia merits treatment when glandular enlargement has been present for more than a year. At this point, the breast tends to become fibrotic and is less likely to resolve spontaneously. Even mild gynecomastia has been found to have significant negative effects on psychosocial well-being.⁴³ If possible, surgical correction before the development of areolar enlargement and skin redundancy is preferable. Ultrasonic liposuction (USL) alone is effective in many patients with grades I and IIA gynecomastia. Mild skin excess will often self-correct after USL. We prefer to use a semicircular, periareolar incision for glandular resection, combined with UDL for grade IIB and grade III gynecomastia. Before the advent of USL, incisions that were used to excise excess skin resulted in unacceptable chest wall scars. USL has allowed increased breast volume reduction without an added scar burden. A USL trochar is introduced through a small stab incision in the proximal anterior axillary line; this provides access to tissue not directly excised through the periareolar incision.^{1,2} Contour irregularities secondary to edema and seroma usually resolve without additional intervention¹ (Fig. 60-12). In patients with persistent skin excess, a circum-areolar incision may be required to resect skin; however, this is only done as a secondary, staged procedure. In patients with very severe glandular enlargement and skin excess, especially with marked enlargement of the areola and ptosis, good results can result with a breast amputation by placing the scar at the inferior edge of the pectoralis major muscle, with free grafting of the nipple-areola complex (Fig. 60-13).

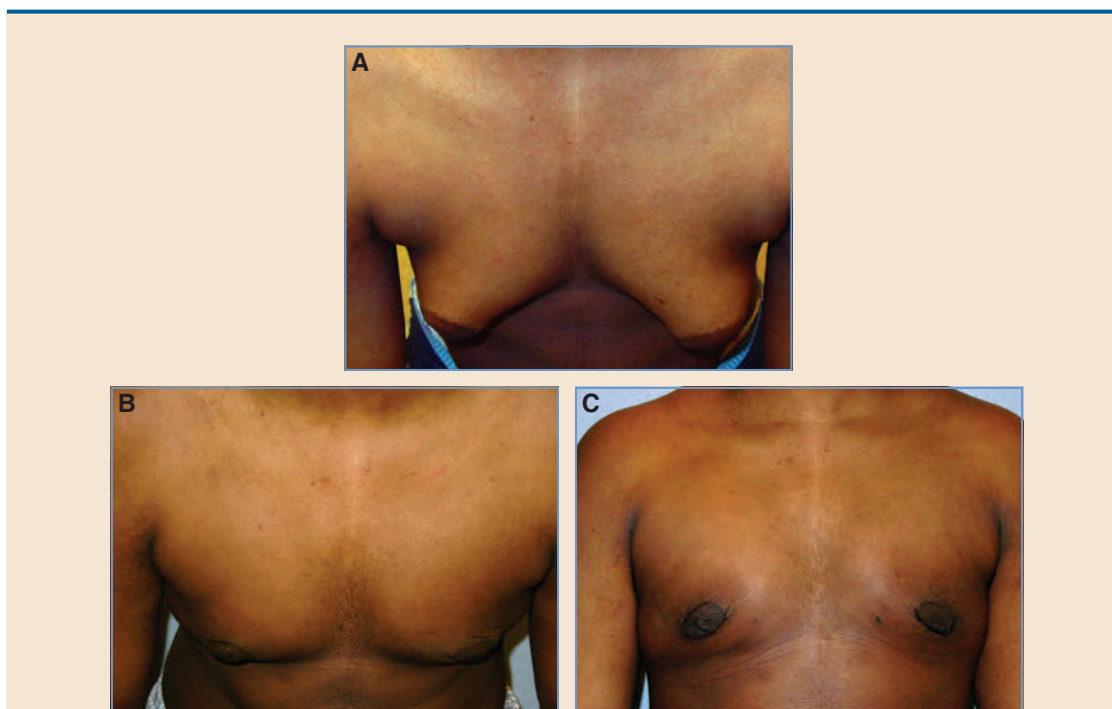


Fig. 60-12 **A**, This adolescent male had gynecomastia. **B** and **C**, He is shown postoperatively after glandular excision with a semicircular, periareolar incision and USL. No skin was excised.



Fig. 60-13 This male patient with gynecomastia underwent a glandular excision with an inframammary approach and free nipple grafting.

Breast Reduction

Patients with macromastia often present with debilitating back, neck, and shoulder pain and have frequently had to curtail athletic activities; breast reduction during adolescence may be warranted for selected cases.^{44,45} A variety of breast reduction techniques can provide good results. We prefer an inferior pedicle technique with a central mound to preserve blood supply and sensory innervation to the nipple. Revisions subsequent to continued breast growth are not usually required. For girls with smaller degrees of hypermastia in whom a vertical reduction technique may be a reasonable option, we prefer to delay surgery until developmental maturity. Often, patients with Poland syndrome or unilateral hypoplasia have an overly large breast on the more normal side.



Fig. 60-14 This patient underwent serial fat grafting for correction of a burn scar deformity. **A** and **B**, Preoperative images. **C** and **D**, She is shown after two sessions of fat grafting. (Courtesy of David A. Billmire, MD, FACS, FAAP.)

Depending on the patient's preference, reduction or mastopexy may be an appropriate symmetry procedure, but this should be delayed until breast growth is complete.

Treatment of Deformational Breast Anomalies

Depending on the deformity, either reduction or augmentation techniques may be required. Burn deformities respond well to tailored reconstructive techniques, including scar release, local flaps, skin grafts, and tissue expansion.^{46,47} Fat grafting may be particularly useful in burned or scarred breasts in restoring volume and releasing tethered scars⁴⁸ (Fig. 60-14). Mastopexy techniques with various pedicles can help to reposition the breast mound or nipple-areola complex. Careful attention to the degree of underlying chest wall asymmetry is important to obtain proper results in patients with scoliosis or pectus deformities.

CONCLUSION

Pediatric breast deformities are relatively common and wide-ranging in presentation. Categorizing these anomalies into the broad groups of hypoplastic, hyperplastic, and deformational facilitates discussion about causes and treatment options. Hypoplastic anomalies require augmentation mammoplasties, whereas hyperplastic breast abnormalities benefit from a variety of reduction

techniques. Autologous fat grafting is a new technique that allows earlier, staged reconstruction and precise correction of volume deficiencies. Careful consideration of surgical timing and the need for staged operations aids in anticipating and optimizing clinical outcomes.

KEY POINTS

- Pediatric breast abnormalities are relatively common.
- Congenital cases can be diagnosed early.
- Surgical treatment is usually delayed until adolescence.
- Pediatric breast abnormalities can be categorized as hypoplastic, hyperplastic, or deformational.
- Hypoplastic abnormalities have the following characteristics:
 - They are less common than hyperplastic abnormalities.
 - They require breast augmentation.
 - Implant augmentation or tissue transfer should be delayed until breast growth is complete, whereas fat grafting can begin as soon as a patient requests correction.
- Hyperplastic abnormalities have the following characteristics:
 - They are more common than hypoplastic abnormalities.
 - They require breast reduction.
 - Symptomatic patients may require surgery before breast development is complete.
- In patients with deformational abnormalities, contralateral symmetry procedures should be performed when breast growth is complete.

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61

Hypospadias

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he word *hypospadias* derives from the Greek words *hypo* (under) and *spadon* (a rent) and describes a condition in which the urethral meatal opening is proximal to the normal orthotopic location of the urethral meatus at the tip of the glans penis. Historically, the correction of hypospadias has been a challenging problem for surgeons. Galen, a physician to the gladiators in Rome, described a partial penectomy, that is, resection of the penis distal to the meatus, as a surgical treatment of this disease.¹ Since then, hypospadias corrective surgery has evolved and has become more sophisticated as surgical techniques and instrumentation have improved. More than 300 procedures for hypospadias repair have been described. Many of these are modifications of techniques promoted by predecessors. This chapter discusses the pertinent anatomy, diagnosis, and classification of hypospadias. The most common current techniques for surgical correction in pediatric urologic and plastic surgery practice are presented.

A host of genetic and environmental factors have been linked to hypospadias, suggesting a multifactorial cause. The genetic basis will be better understood as genetic testing becomes more common and several potentially influential genes and polymorphisms are further evaluated² (Box 61-1).

Endocrine dysfunction, including estrogen effects, a failure of androgen stimulation caused by incorrect timing of hormone production, an insufficient androgen supply, or end-organ receptor malfunction, is thought to be the most common underlying pathway toward the development of hypospadias. The incidence of hypospadias varies with geographic region and appears to be increasing. Although the increase was initially thought to be caused by an increased detection of minor forms of hypospadias, Canon et al³ recently described an increase in the prevalence of all forms (coronal/distal, midpenile/subcoronal, and perineal/proximal penile) of hypospadias in Arkansas. The overall rates of hypospadias increased from 6.69 per 1000 (from 1998 to 2002) to

Box 61-1 Genes Investigated as Possible Causes of Hypospadias

<i>5A2</i>	<i>ESR1</i>	<i>GRID1</i>	<i>PPARGC1B</i>
<i>AR</i>	<i>ESR2</i>	<i>HOXA4</i>	<i>SF1</i>
<i>ATF3</i>	<i>FGF8</i>	<i>HOXB6</i>	<i>SHH</i>
<i>BMP7</i>	<i>FGFR2</i>	<i>HSD17B3</i>	<i>SRD</i>
<i>CYP11A1</i>	<i>GLI1</i>	<i>HSD3b2</i>	<i>SRD5A2</i>
<i>DGKK</i>	<i>GLI3</i>	<i>MAMLD1</i>	<i>WT1</i>

8.10 per 1000 (from 2003 to 2007).³ No clear reasons for these increases are evident; however, increased reporting of hypospadias, changes in genetic susceptibility, increased maternal age, improved survival of low-birth-weight infants, and environmental exposures may have an impact.³

PERTINENT NORMAL AND ABNORMAL ANATOMY

There are important functional and cosmetic reasons for considering surgical correction of hypospadias. Relocation of the urethral meatus to the tip of the penis facilitates a straight urinary stream, allowing a child to stand to void.⁴ It helps to improve semen deposition, which can affect fertility potential. This is most significant for the more severe forms of hypospadias. The associated chordee (an abnormal curvature of the penis) is corrected by orthoplasty in which the penis is straightened, improving sexual function and vaginal penetration later in life.⁵ Although long-term results have shown that patients with corrected hypospadias are more likely to be less satisfied with their penile appearance, they are as sexually active as controls.⁴ With hypospadias correction, the cosmetic appearance of a child's genitals resembles that of a normal circumcised penis. In selected cases, the foreskin can be preserved if an uncircumcised appearance is desired.⁶

EMBRYOLOGY

Hypospadias occurs when the process of normal urethral development is interrupted, and the urethral meatus does not reach its orthotopic location at the tip of the glans. The formation of the urethra begins at approximately 4 weeks of gestation, when the urorectal fold forms and divides the cloacal membrane into an anterior urogenital sinus and a posterior rectum.⁷ The urethral plate can be seen as a thickening of the anterior wall of the endodermal cloaca.⁸ Simultaneously, paired swellings anterolateral to the cloacal membrane become visible and fuse into a midline genital tubercle at 6 weeks of gestation.⁷ The lateral mesoderm forms urethral and genital folds. The urogenital sinus elongates onto the ventral surface of the phallus to the level of the corona and forms the urethral groove. The walls of the urogenital sinus join to form the endodermal urethral plate on the floor of the urethral groove. The groove deepens and forms the anterior urethra. Under the influence of testosterone, the fetal phallus elongates, and the urethral groove extends to the level of the corona. The urethral folds unite at the midline, closing the urethra and

forming the median raphe of the scrotum and penis. The glans closes over the urethral groove, and the underlying epithelial cells tubularize and form the glandular urethra, which grows proximally to join the penile urethra.⁹ The preputial skin forms as a ridge that gradually grows to enclose the glans circumferentially. A defect associated with hypospadias is deficient foreskin ventrally, resulting in a dorsal hood.⁷

Chordee is frequently associated with hypospadias. Glenister,⁸ in his extensive anatomic study of urethral plate and phallic development in fetuses, found that penile curvature is a normal stage in embryologic development; this finding suggests that chordee may result from an arrest of genital development at an early embryologic stage. In hypospadias, the severity of chordee parallels the severity of the defect of the urethral plate. Thus the more proximal forms of hypospadias are associated with more severe chordee.

CLASSIFICATION

Traditionally, hypospadias has been classified based solely on the location of the hypospadiac urethral meatus, with the following general categories: distal shaft (anterior urethra), midshaft (midurethra), and proximal shaft (posterior urethra)¹⁰ (Fig. 61-1). Recently, a new grading schema was proposed qualitatively describing not only the location of the meatus but also the character of the glans and chordee of the shaft, and it has been shown to correlate with the risk of postoperative complications¹¹ (Table 61-1).

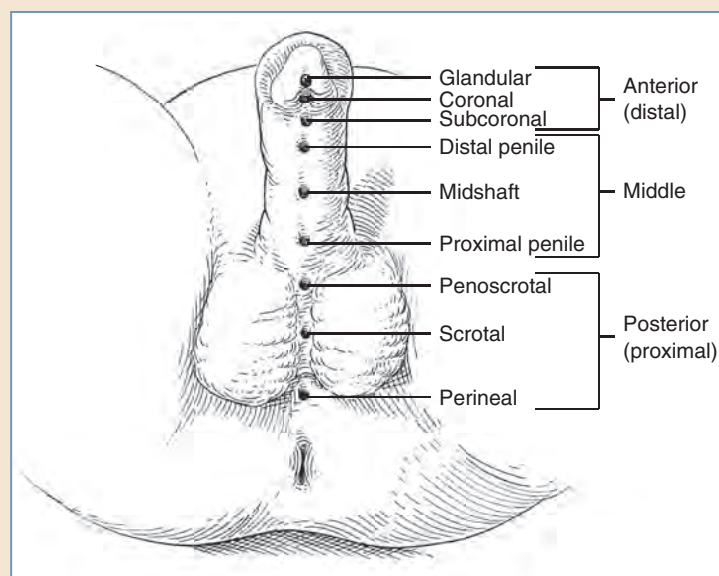


Fig. 61-1 Hypospadias is traditionally classified by the anatomic location of the urethral meatus.

Table 61-1 Glans-Urethral Meatus-Shaft* Scoring Criteria for Hypospadias

Glans Score (G)	
Glans good size, healthy urethral plate, deeply grooved	1
Glans adequate size, adequate urethral plate, grooved	2
Glans small in size, urethral plate narrow, some fibrosis or flat	3
Glans very small, urethral plate indistinct, very narrow or flat	4
Meatus Score (M)	
Glanular	1
Coronal sulcus	2
Mid or distal shaft	3
Proximal shaft or penoscrotal	4
Shaft Score (S)	
No chordee	1
Mild (<30 degrees) chordee	2
Moderate (30-60 degrees) chordee	3
Severe (>60 degrees) chordee	4

*This scoring system, also known as the *GMS scoring system*, incorporates traditional meatus scoring with new glans and shaft scoring.

PREOPERATIVE AND PRETREATMENT ASSESSMENT

The diagnosis of hypospadias is usually made based on the initial newborn examination. However, with improvements in prenatal ultrasound imaging, hypospadias can be diagnosed antenatally because of changes in the distal morphology of the penis, including small lateral folds corresponding to the dorsal hood, visible chordee, and, rarely, an abnormal fetal urinary stream.¹² More minor forms of hypospadias might not be diagnosed until adulthood. Regardless of the age of presentation of a patient with hypospadias, a full prenatal and birth history should be obtained to identify potential hormonal and familial factors and to evaluate other associated conditions. To help determine whether a patient has functional deficits, the parents are asked about the direction of their child's urinary stream and the presence of curvature of the erect or flaccid penis.

A physical examination includes assessment of the following: the degree of chordee, the size and configuration of the glans, the integrity of the ventral penile skin, the quality and depth of the urethral plate, the amount of dorsal hood foreskin, and, most important, the location of the urethral meatus. In patients with significant chordee and a disproportionately distal hypospadiac meatus, the true defect is probably more severe than the meatus location suggests. In these cases the skin overlying the urethra proximal to the hypospadiac meatus is usually overly thin and associated with a poorly formed urethra, and the true level of the hypospadiac defect is much more proximal.

A careful scrotal and inguinal examination completes the genitourinary examination. Hypospadias is associated with other genital abnormalities such as cryptorchidism (8%), hydrocele (16%), and inguinal hernia (8%).¹³ Associated malformations are more likely to occur in patients with more severe hypospadias. Patients with proximal hypospadias can have associated renal

anomalies, which can be evaluated by ultrasound imaging; however, routine imaging of the urinary tract in all cases of hypospadias is not necessary.

It is important to always keep in mind the possibility of a disorder of sex development (DSD) when evaluating a child with hypospadias. A wide range of rates (4.2% to 100%) has been reported for chromosomal anomalies in patients with both cryptorchidism and hypospadias.¹⁴⁻¹⁶ This wide range is probably the result of variances in the severity of hypospadias and cryptorchidism. In patients with proximal hypospadias, the incidence of chromosomal abnormalities is higher, and it is three times more likely if one or both of the gonads are nonpalpable.^{15,17} Conclusions about an appropriate workup for these patients are controversial based on the wide range in incidence of DSD.¹⁷ The most common DSD in these cases is mixed gonadal dysgenesis, a disorder characterized by a mosaic karyotype, a unilateral streak gonad, a unilateral testicle (frequently undescended), and persistent müllerian structures. In general, a good rule of thumb is to perform a urologic and an endocrine workup in all patients with hypospadias and a nonpalpable testicle, because some conditions may have serious consequences if not identified early. A classic example of this is congenital adrenal hyperplasia caused by 21-hydroxylase deficiency. These patients can die of salt wasting caused by ineffective glucocorticoid and mineralocorticoid synthesis. The phenotype of these patients depends on the severity of the associated excess androgen production. Those with a normal female karyotype have nonpalpable gonads and a phenotype ranging from severe hypospadias to a fully formed phallus.

NONSURGICAL AND SURGICAL TREATMENT OPTIONS

As mentioned previously, relocating the urethral meatus to the tip of the glans facilitates the production of a straight urinary stream and the deposition of ejaculate within the vaginal fornix. Correcting the associated chordee improves the dynamics of vaginal penetration during intercourse and allows the patient to stand while voiding. Finally, approximating the penile appearance to other males is an important cosmetic factor that can have psychosocial implications later in life and should be addressed when counseling a family regarding the need for hypospadias surgery. In the absence of cosmetic or significant functional deficits, some mild forms of hypospadias do not necessarily “need” surgical correction. Other than issues with sexual function secondary to chordee, a recent survey showed that adult men with mild, untreated hypospadias had no deficits in their fertility, urinary function, quality of life, or satisfaction with the cosmetic appearance of their penis, compared with controls.⁵

Hypospadias surgery has no contraindications other than medical fitness to undergo general anesthesia. Proper, informed consent can only be obtained after thorough counseling regarding the risks and benefits of corrective surgery and the expected functional and cosmetic results of surgery. Hypospadias is a surgical disease, and presently no specific nonsurgical treatments are available.

PREOPERATIVE AND PRETREATMENT PLANNING

Timing of Surgery

The optimal timing of hypospadias repair is controversial. Most hypospadias surgeons think that the ideal patient age for an elective repair of hypospadias is 6 to 12 months of age.¹⁸ By 4 to 5 months of age, the anesthetic risk for infants has decreased, especially when the procedure is performed by an anesthesiologist with pediatric experience, and proper monitoring techniques are used. Studies have shown that the psychological effect of surgery on children 1 to 3 years of age is increased.¹⁸ Therefore emotional distress from genital surgery is probably lower in younger patients. The size and quality of the genital tissues after 6 months of age are usually adequate for

hypospadias repair. Conversely, a recent study reported no difference in long-term psychological adjustment, health-related quality of life, and surgical outcomes in boys who had undergone surgery before or after 18 months.¹⁹ Bush et al²⁰ and Snodgrass et al²¹ compared the complications of hypospadias repair in older children and adults after tubularized incised plate repair (TIP) urethroplasty with those in younger children and found no difference.

In one survey, 78% of pediatric urologists used preoperative hormonal stimulation in patients with proximal hypospadias and in patients with a microphallus and/or a reduced glans circumference or urethral plate.²² Androgen stimulation in the form of human chorionic gonadotropin injection, parenteral testosterone injection, dihydrotestosterone transdermal gel, and topical testosterone cream (2%) increases the size of the penis and the vascularity of the tissues. However, preoperative hormonal stimulation has potential downsides, with basic science research showing a negative impact of androgens on wound healing, potential exposure of the parents to topical hormonal therapies, patient pubic hair growth, aggressive behavior, and potential patient linear height and bone age changes.²³ The current literature does not report the risk-benefit ratio of preoperative hormonal stimulation. In a recent systematic review, Wright et al²³ showed a non-significant, increased risk of surgical complications in patients who underwent preoperative hormonal stimulation (OR 1.67; CI 0.96-2.91).

In conclusion, current indications for preoperative hormonal stimulation include children with proximal hypospadias, microphallus, and/or reduced glans width or reduced urethral plate. In most cases of routine distal or midhypospadias, hormonal stimulation is not indicated. In our practice, we utilize a 2- to 3-week period of hormonal stimulation to take advantage of the improved vascularity and tissue growth, and we stop the medication 1 week before surgery, thereby theoretically limiting the acute changes related to hormonal stimulation and its negative effects on wound healing.

SURGICAL TECHNIQUE

The surgery is performed with the child under general anesthesia. At many institutions, the anesthesiologist or the surgeon routinely performs a caudal epidural anesthetic block for pain control perioperatively. Alternatively, a local penile nerve block is performed rather than a caudal block. This prevents the possible penile engorgement seen with caudal blocks and potentially decreases the risk of postoperative complications, as Kundra et al²⁴ reported in a randomized trial. For the duration of the procedure, the child is positioned supine on the operating table, with the legs slightly abducted and flexed at the knees. All pressure points are carefully padded. A routine cystourethroscopy is not generally performed simultaneously unless indicated for other reasons.

To ensure precise placement of sutures and gentle handling of fine tissues, optical magnification during hypospadias surgery is the rule. Although most surgeons find that surgical loupes with magnification of 2.5× or higher are satisfactory, an operating microscope can be used. Intraoperative bleeding during hypospadias surgery can be minimized by intermittent use of a small tourniquet applied to the base of the penile shaft. At our institution, we have found that a thin rubber band and a hemostat (clamped over a dental roll that is positioned ventrally over the proximal urethra for protection from inappropriate compression from the rubber band) are very useful for this purpose. Alternatively, some surgeons prefer to inject the tissues with epinephrine solution (1:200,000) to prevent excessive bleeding during surgery in the well-vascularized glans and penis.

The specific surgical technique used for hypospadias correction depends on several important factors that surgeons must evaluate preoperatively and intraoperatively. These include the location of the hypospadiac meatus; the severity of the chordee; the width, depth, and health of the urethral plate; the quality of the penile shaft skin; and the presence and amount of dorsal hood tissue. The principal techniques for distal, midshaft, and proximal forms of hypospadias are discussed and illustrated in this section.

In the past, surgeons preferred to stage hypospadias and associated chordee correction with two or more procedures. More recent technical advances have allowed single-stage repairs in most typical cases of hypospadias. Presently, single-stage repairs are the benchmark for distal and most midshaft hypospadias. For patients with proximal hypospadias, especially those associated with severe chordee, whether a single-stage or a two-stage procedure is more appropriate is controversial. The pros and cons of each of these approaches are discussed in the following sections.

Distal Hypospadias

Distal hypospadias is defined as a meatus proximal to the coronal sulcus of the glans (subcoronal), at the sulcus (coronal), or distal on the glans. It is not located at the orthotopic position on the tip (glanular). Usually, the meatus is subcoronal, the associated chordee is mild, and the major goal of surgery is to produce a functional distal urethra. Currently, the most commonly used surgical procedures to correct distal hypospadias include a meatoplasty and glanuloplasty (MAGPI); a flip-flap repair, also known as a *Mathieu repair*; and a tubularized incised plate (TIP) urethroplasty, also known as a *Snodgrass repair*.

Duckett²⁵ originally described the MAGPI procedure¹⁰ for correcting distal hypospadias without chordee (Fig. 61-2). Essentially, the hypospadiac meatus is incised in a longitudinal fashion and closed transversely, that is, in the fashion of Heineke-Mikulicz. The penile skin is degloved circumferentially, and incisions are made into the glans ventrally to create glans flaps. The flaps are brought together proximal to the neomeatus, which has the effect of advancing the urethra and neomeatus distally. Finally, penile skin is reapproximated to the glans collar. An advantage of this technique is that no formal reconstruction of a neourethra is necessary; thus the likelihood of a fistula formation is extremely low. In a few, carefully selected patients, a MAGPI repair provides a very nice functional result. However, in most patients the cosmetic result is less than satisfactory, with a flattened glans and meatal retrusion. Therefore, at most centers, this is not the preferred method for repairing routine cases of distal hypospadias.

The flip-flap technique (or Mathieu technique)²⁶ uses a midline meatal-based flap of skin on the ventral penile shaft to form the anterior wall of the neourethra (Fig. 61-3). The glans is incised

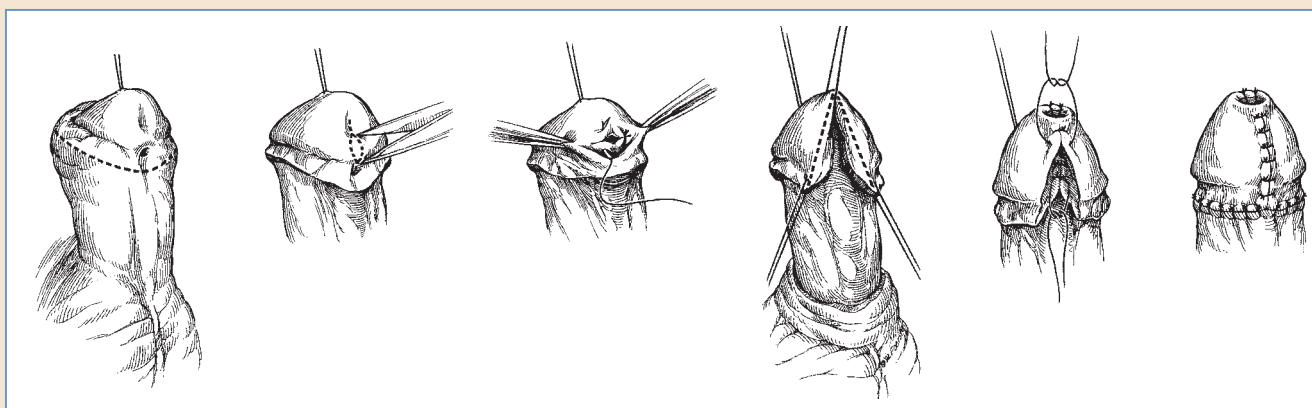


Fig. 61-2 The MAGPI procedure. A circumcising incision is made, the urethral meatus is incised longitudinally and closed transversely, advancing it distally in the manner of Heineke-Mikulicz. The glans flaps are reapproximated proximally to complete the repair.

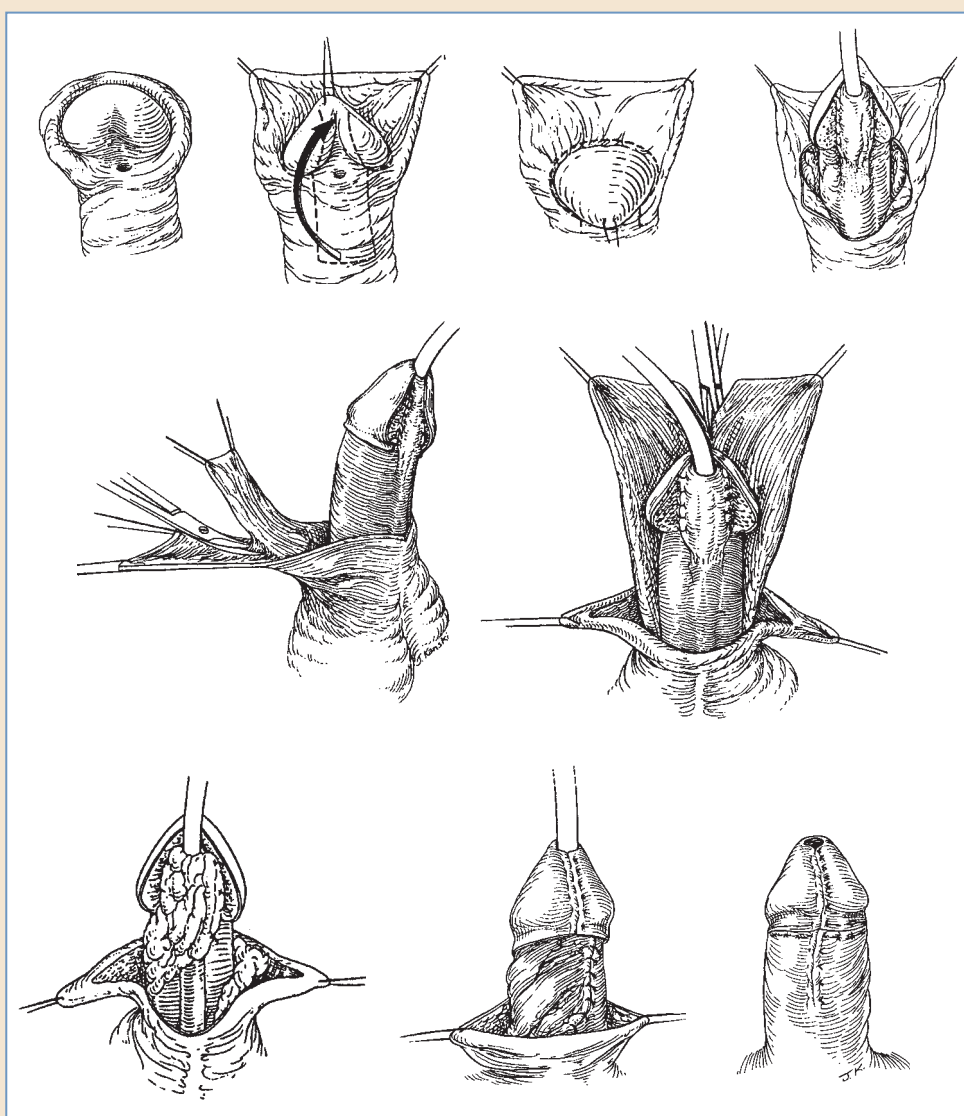


Fig. 61-3 The flip-flap (or Mathieu) repair. A midline skin flap is created from the ventral penile skin and rotated to create the neourethra distally. A vascularized dorsal dartos flap is used to cover the repair, and the remaining skin is used for penile shaft coverage.

laterally on either side of the urethral plate, and the flap is pivoted distally to create the neourethral tube. The suture lines are covered with a vascularized subcutaneous flap harvested from the preputial and shaft skin before the glanuloplasty. The advantages of the Mathieu technique are that it is not technically demanding, it is highly successful, and the suture lines do not overlap, which decreases the risk for fistula formation. Disadvantages include the variability of the blood supply to the ventral flap, which can potentially cause flap necrosis or fistula formation. Another disadvantage is the cosmetic appearance of the neomeatus, described as rounded and horizontal or like a “fish mouth,” as opposed to the desired slitlike meatus.²⁷

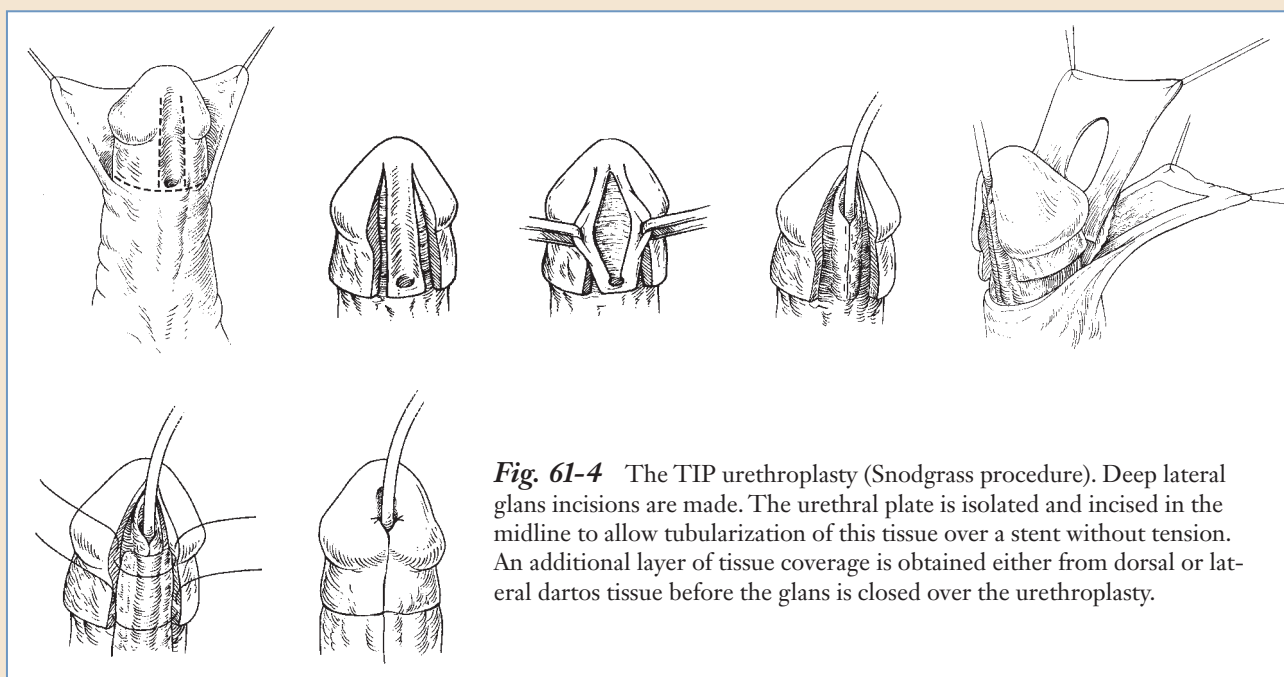


Fig. 61-4 The TIP urethroplasty (Snodgrass procedure). Deep lateral glans incisions are made. The urethral plate is isolated and incised in the midline to allow tubularization of this tissue over a stent without tension. An additional layer of tissue coverage is obtained either from dorsal or lateral dartos tissue before the glans is closed over the urethroplasty.

TIP urethroplasty is based on a modification of Thiersch-Duplay urethroplasty, which incorporates incising the urethral plate and reconstructing the urethra by tubularization of the urethral plate in situ. Rich et al²⁸ originally described incising the urethral plate in a variety of repairs. Snodgrass²⁹ popularized the longitudinal incision of the urethral plate specifically in association with the Thiersch-Duplay repair. This incision of the urethral plate avoids the potential problems with the Thiersch-Duplay urethroplasty, specifically, a glans that is disproportionately small in relation to the penile shaft and a glans closure that is too tight. This theoretically decreases the risk for dehiscence and fistula. Incision of the plate allows a tension-free anastomosis and decreases the need to incorporate surrounding glans tissue in the urethral reconstruction. Reclosure of the urethral plate incision postoperatively and urethral strictures are a concern. However, basic science research demonstrated that the area of incision reepithelializes quickly, and a meta-analysis comparing the flip-flap to TIP urethroplasty showed no significant differences in fistula development, meatal stenosis, or wound dehiscence.^{30,31}

A TIP urethroplasty, also known as the Snodgrass procedure²⁹ (Fig. 61-4), involves tubularization of the urethral plate using flaps created from parallel, longitudinal incisions in the glans; this maneuver creates glans wings for closure. The Snodgrass modification consists of a deep midline incision in the urethral plate, from the hypospadiac meatus to the tip of the glans. The urethra is closed ventrally in two layers, and the epithelium is inverted. The urethral meatus has a normal, vertical, slitlike appearance. One of the potential drawbacks to this procedure is that all suture lines overlap in the midline. Therefore many surgeons routinely cover the repair with a vascular, deepithelialized flap from either the prepuce or lateral ventral tissue.³² This practice was recently debated after a prospective randomized trial showed no difference in complications between the use of a dartos flap and a flapless TIP repair³³ (Fig. 61-5). Regardless of flap utilization, the TIP procedure provides optimal functional and cosmetic results, and it is our procedure of choice for repairing most cases of distal hypospadias.

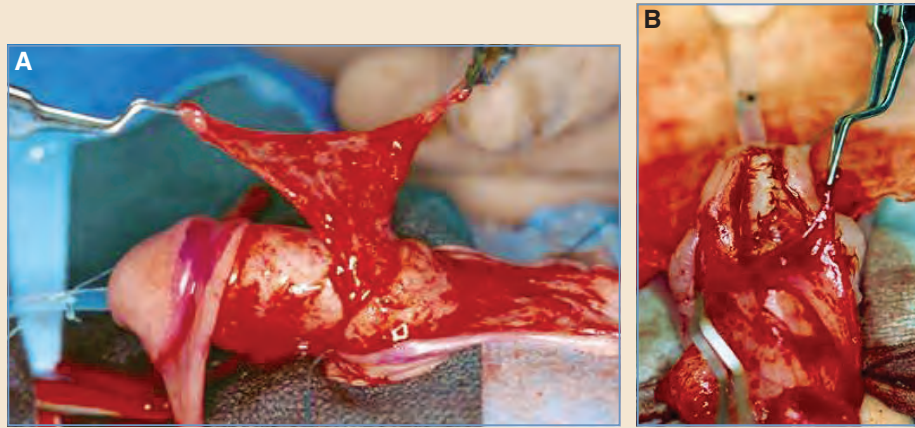


Fig. 61-5 Harvesting a vascularized flap. **A**, A vascularized dartos flap is harvested from the subcutaneous tissue of the dorsal penile skin and preputial skin. **B**, The flap is rotated on its pedicle to provide ventral coverage.

Of the multiple surgical options available to correct distal hypospadias, the optimal procedure for a particular patient is operator dependent and individualized to the patient's particular anatomy. Currently, the trend is toward using a TIP urethroplasty as the operation of choice for distal hypospadias, primarily because it provides excellent overall cosmetic and functional results.³⁴

Foreskin preservation, or preputial reconstruction, is possible with hypospadias repair. The role of preputial reconstruction in hypospadias is controversial and currently debated. Some surgeons are concerned that preputial reconstruction may increase complications because of alterations in the blood supply to the urethral reconstruction and because of an inadequate amount of skin for reconstruction, which increases the risk of phimosis. The technique for foreskin reconstruction involves layer-by-layer reconstruction. An incision is made along the lateral edges of the hooded foreskin, and the prepuce is divided into three layers. The foreskin is sutured in the midline in three layers; the first two layers (the inner prepuce and dartos fascia) are approximated in a continuous fashion and the outermost layer (outer prepuce) in an interrupted fashion.³⁵ The reported complication rates for preputioplasty vary significantly. However, in a recent report in which preputioplasty was offered only to patients with favorable anatomy, only 3.9% required a secondary circumcision.³⁵

The decision to preserve the foreskin should be made by the parents. Informed consent is obtained after parents are provided with objective data regarding the potential benefits of preputioplasty and complication rates, which vary depending on the severity of the hypospadias and the foreskin anatomy. This is weighed, along with the parents' cultural/personal desires for an uncircumcised appearance, to determine the appropriate surgical intervention.

Midshaft Hypospadias

Hypospadias on the midshaft portion of the penis is usually associated with more significant chordee than distal hypospadias. Therefore correction of chordee is a much more significant component of the surgical procedure. (This portion of the procedure is discussed in detail in the following section on proximal hypospadias). TIP urethroplasty is still an excellent option for midshaft hypospadias when the integrity of the urethral plate is good. In some cases of more severe

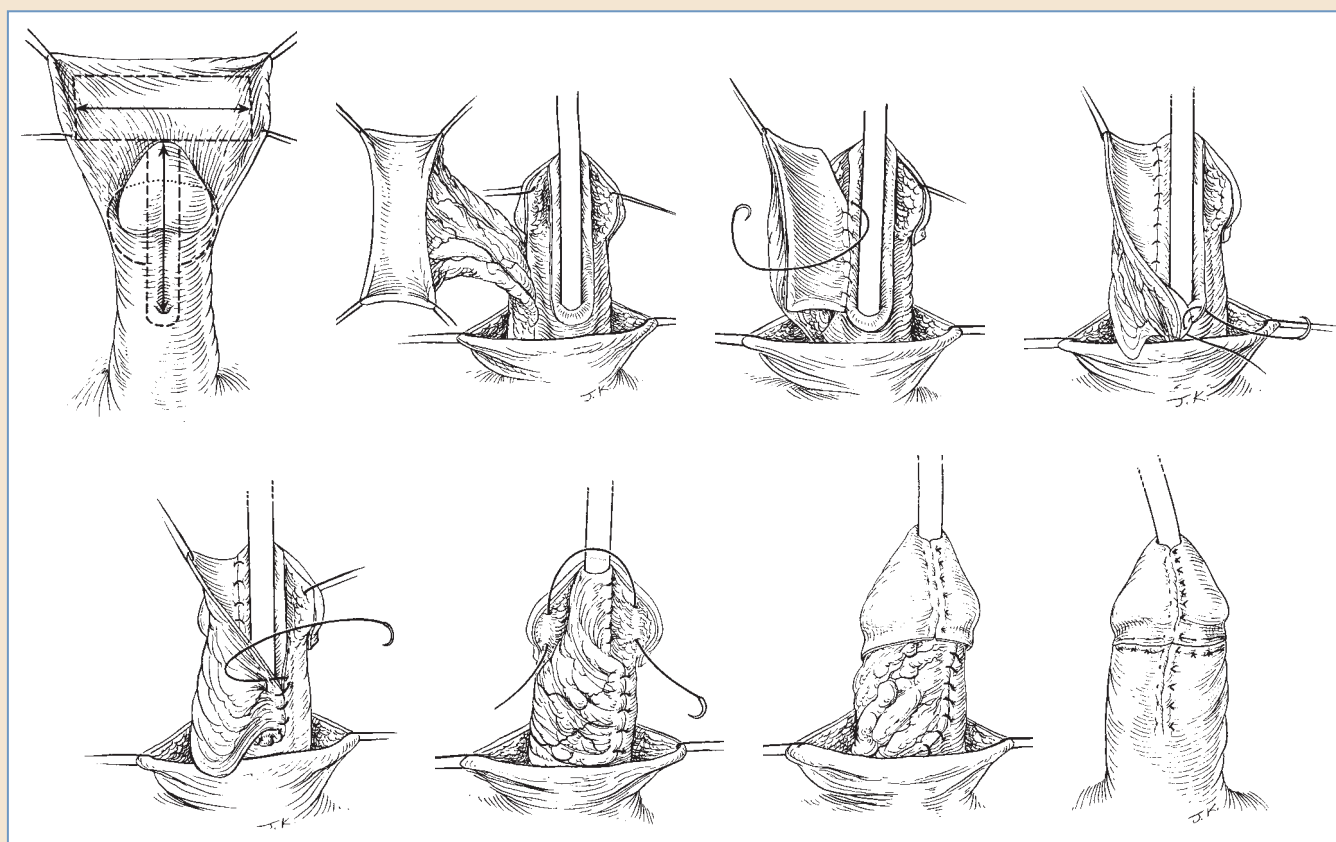


Fig. 61-6 The onlay island flap. A vascularized island of skin with a pedicle is harvested from the inner preputial skin and anastomosed to the urethral plate with two lateral suture lines. The glans is reapproximated over the closure, and the penile skin is closed to complete the repair.

midshaft hypospadias, the urethral plate is either too narrow or not supple enough for tubularization without tension, even after a deep incision is made in the base. In such cases, alternative techniques should be considered. However, patients with an adequate urethral plate achieve good functional and cosmetic results with a low rate of complications.³⁶

The other common technique employed for midshaft hypospadias²⁶ is an onlay island flap urethroplasty (Fig. 61-6). The urethral plate is preserved as the posterior urethral wall. The anterior urethral wall is created from a strip of skin usually obtained from the inner preputial skin of the foreskin, harvested with its native blood supply. A vascularized flap of penile skin may alternatively be used. The flap is rotated ventrally and anastomosed onto the urethral plate with two lateral running suture lines. Glans wing flaps are created and closed in the midline. The penile skin is closed primarily, or the ventral penis is resurfaced using skin flaps from the dorsal prepuce (Byars flaps). The onlay island flap has been shown to have good cosmetic results; however, it increases the risk for a urethral diverticulum. This is probably the result of poor tissue support for the anterior neourethral wall, which may promote outward ballooning of the neourethra.³⁷ In a recent systematic review and meta-analysis comparing the onlay island flap procedure with TIP urethroplasty, the onlay island flap repair had relatively few complications, especially in patients with little or no chordee.³⁷

Proximal Hypospadias

When the hypospadiac meatus is in a more proximal location (penoscrotal, scrotal, or perineal), corrective surgery is less straightforward and more challenging. Not only is the length of urethra requiring reconstruction greater, but also these patients generally have a greater degree of chordee, and the urethral plate may be fibrotic, narrow, or nonexistent. Depending on a patient's characteristics, the hypospadias can be repaired in several surgeries (staged) or all at once (single-stage). A single-stage approach corrects chordee and builds a urethral tube extending to the glans in one surgery. A staged repair corrects the chordee and provides a foundation of tissue on the urethral plate that will be used to complete the repair 6 to 12 months later.

Adequate correction of the chordee (orthoplasty) is the first step in staged and single-stage repair. The initial approach to orthoplasty is to deglove the penile skin and aggressively dissect and excise abnormal fibrous chordee tissue from the corporal bodies. In some cases this dissection alone is sufficient to straighten the penile shaft. To assess the degree of residual chordee after this dissection, an artificial erection is created with a tourniquet, as described previously for control of blood loss. Saline solution is injected into one of the corporal bodies with a butterfly needle. Persistent chordee (greater than 20 to 30 degrees) is corrected with one of several approaches. Nesbit³⁸ described removal of small transverse strips of tissue from the dorsal surface to equalize the disparity between the surfaces (Fig. 61-7). A modification of this technique closes these longitudinal incisions horizontally with plicating sutures in the manner of Heineke-Mikulicz.³⁹ Baskin et al⁴⁰ performed anatomic studies demonstrating that the distribution of the dorsal neurovascular bundle is more laterally diffuse than previously thought, which suggests that Nesbit plication techniques could injure nerves inadvertently. He therefore proposed a dorsal plication in which a permanent plication suture or sutures are placed in the midline between the neurovascular bundles. This has proved to be an effective method of correcting moderate degrees of residual chordee. If adequate correction of chordee is possible with dorsal plication techniques,

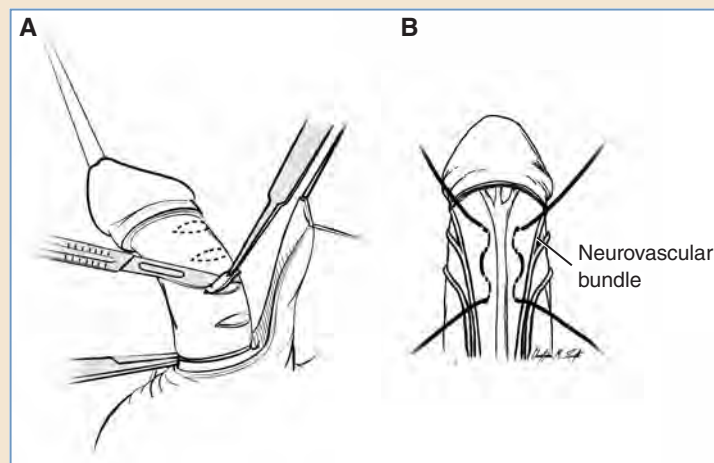


Fig. 61-7 Chordee repair. **A**, The Nesbit tuck procedure. Vertical elliptical segments of tunica albuginea are removed from the longer, more convex portion of the penile shaft to correct the chordee. **B**, Dorsal plication. The tunica albuginea is sutured lateral to the dorsal vein and repeated as necessary to correct the chordee.

then a single-stage procedure can be considered, and the urethra can be reconstructed with one of several techniques.

Several options are available for urethral reconstruction. As mentioned previously, a TIP procedure is an option when the urethral plate is soft and supple. When the urethral plate is present but not adequate for tubularization, then an onlay flap can be used. For cases in which the urethral plate is too deficient to use with onlay flap techniques, a tubular vascularized flap can be used. Duckett^{25,41} first introduced such a flap—the transverse preputial island flap (Fig. 61-8). A

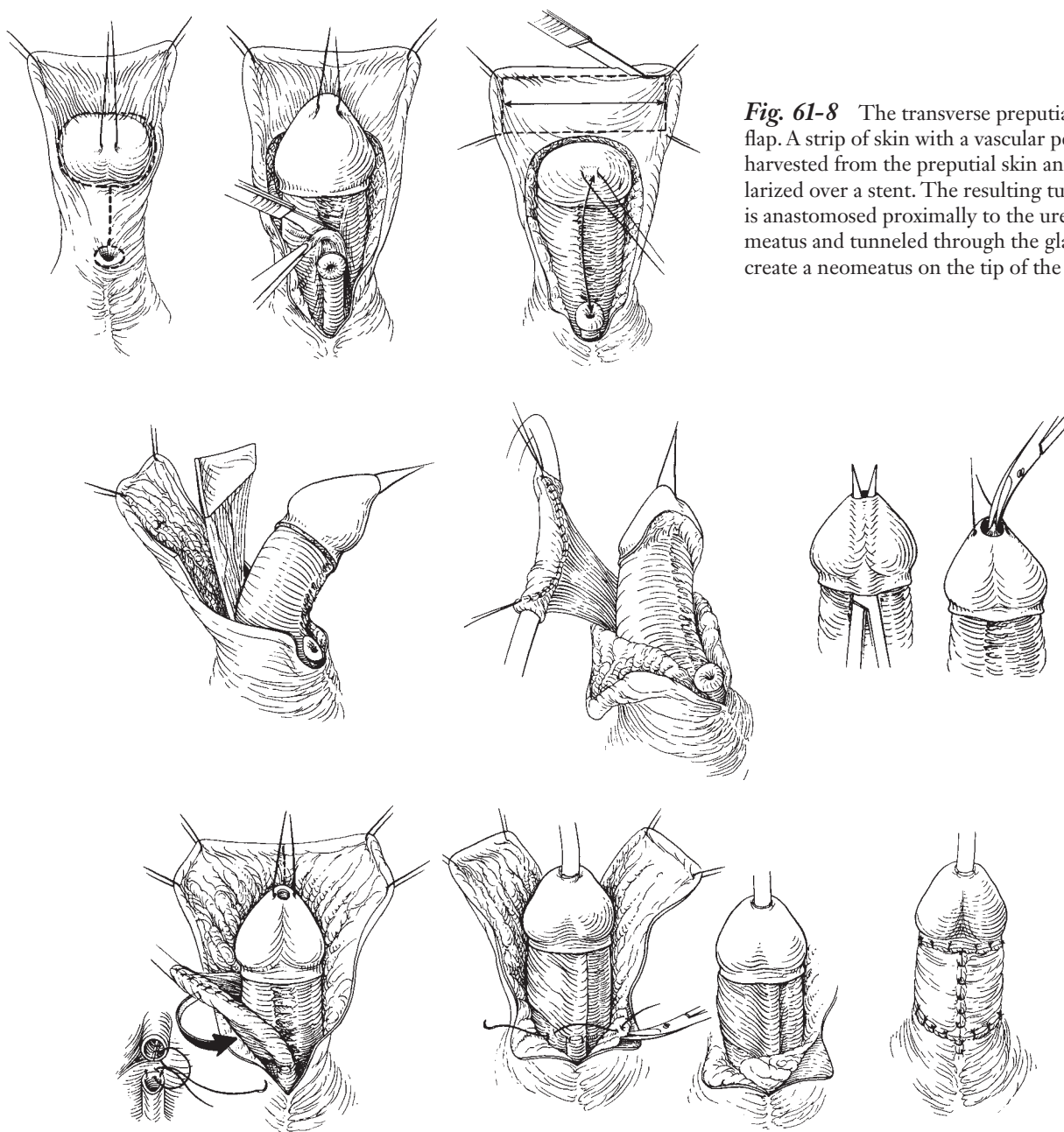


Fig. 61-8 The transverse preputial island flap. A strip of skin with a vascular pedicle is harvested from the preputial skin and tubularized over a stent. The resulting tube flap is anastomosed proximally to the urethral meatus and tunneled through the glans to create a neomeatus on the tip of the penis.

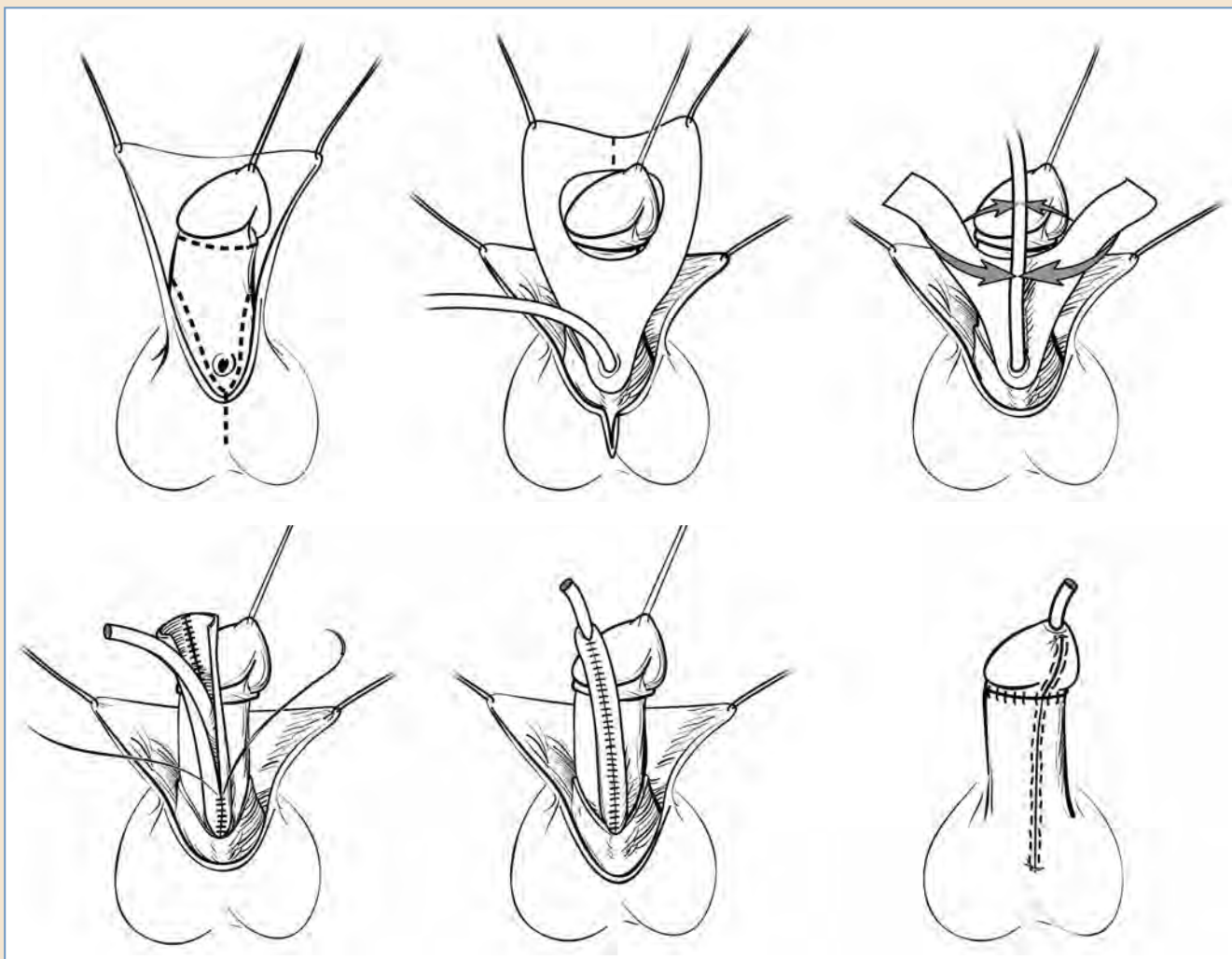


Fig. 61-9 The Koyanagi repair. Yolk-shaped incisions on the parametarial penile tissues facilitate penile degloving. The chordee is corrected, and a tubularized neourethra is created using the ventral and inner preputial skin and an inverting running suture. Distal skin flaps are trimmed as necessary, and the ventral aspect of the penis is resurfaced with Byars flaps.

vascularized strip of dorsal preputial skin is tubularized over a catheter to create a neourethra between the proximal hypospadiac meatus and the tip of the glans. The width of the flap should be approximately 15 mm to roll a tube of adequate diameter. Within the glans, the tube can either be placed between flaps of glans tissue that have been created following a deep incision in the glans, or it can be tunneled through the central portion of the glans to create a neomeatus at the tip.

Koyanagi et al⁴² developed an alternative approach to a single-stage repair of hypospadias that uses flap and tubularization techniques to form the urethra. After the chordee is adequately corrected, a meatal-based preputial flap is used (Fig. 61-9). This repair is referred to as a *manta-wing flap procedure* because of the appearance of the skin flaps used to form the urethra. Regardless of the type of urethroplasty performed, covering the repair with a vascularized flap is essential

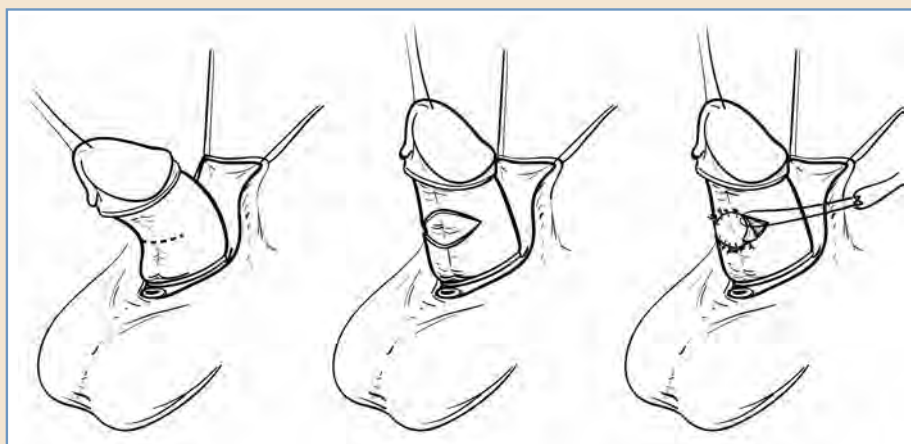


Fig. 61-10 Corporal body grafting. A diamond-shaped wedge of corporal body tissue is removed ventrally and grafted with replacement material to correct severe chordee.

to reduce the risk of breakdown and fistula formation. Similar to correction of midshaft hypospadias, this tissue can be harvested from the surrounding dartos layer of the penis or from the tunica vaginalis, which is harvested from the spermatic cord and the associated testis.⁴³

When significant chordee cannot be appropriately corrected with dorsal plication techniques, then a staged approach is performed. Dividing the urethral plate at the area of maximum curvature and dissecting it away from the corporal bodies allows resection of all remaining fibrotic and dysgenetic tissue contributing to the chordee. In some cases this maneuver sufficiently reduces the chordee to allow correction with a dorsal plication. However, in most cases complete correction of the residual chordee requires corporal body grafting (Fig. 61-10). A deep ventral incision at the area of maximal curvature is made within the corporal bodies, extending to the lateral border of the dorsal neurovascular bundle on each side. This results in a diamond-shaped defect that can be grafted with either a free graft of dermis (from a non-hair-bearing portion of the body), small intestinal submucosa, or tunica vaginalis.⁴⁴⁻⁴⁶ Small intestinal submucosa, a xenograft material, has proved useful for corporal body grafting. It is an off-the-shelf material that requires no donor tissue, facilitates corporal body regeneration, and is as effective as donor tissue from the host.⁴⁶

Once corporal body grafting is completed, skin flaps are rotated ventrally to create a new urethral plate that will be tubularized into a neourethra at the second stage of repair. Different types of flaps can be used for ventral penile skin coverage. The skin is typically made up of a combination of penile shaft and/or preputial skin. Regardless of the type of skin used, adequately preserving the blood supply to the flap and tailoring it along the ventral penis to create a flat plate of skin without redundancy are critical. This facilitates tubularization in the second stage. Skin flaps are also useful for resurfacing the ventral aspect of the glans when the native glandular urethral plate is inadequate.^{47,48} Six to twelve months later, at the second stage, a neourethra is created by incising a 15 mm strip of tissue at the ventral midline, which is then tubularized to the tip of the glans. The lateral skin edges are mobilized, and the remaining tissue is closed over the repair in at least two layers.⁴⁸ Cheng et al⁴⁹ proposed a modification of the traditional staged repair in which the urethral plate is divided more proximally at the level of the hypospadiac meatus to preserve the native urethral plate. The preserved urethral plate can be used for tubularization of the distal

neourethra within the glans and distal shaft using the TIP technique at either the first or second stage⁴⁹ (Fig. 61-11). This modification appears to lower the fistula rate and overall complication rate related to the formation of a neourethra. The glans has a more normal appearance with this technique. This modification is appropriate only in cases in which the native urethral plate in the glans is adequate for primary closure. When the native urethral plate in the glans is inadequate, the distal urethra can be repaired with skin flaps, with good cosmetic results.⁵⁰

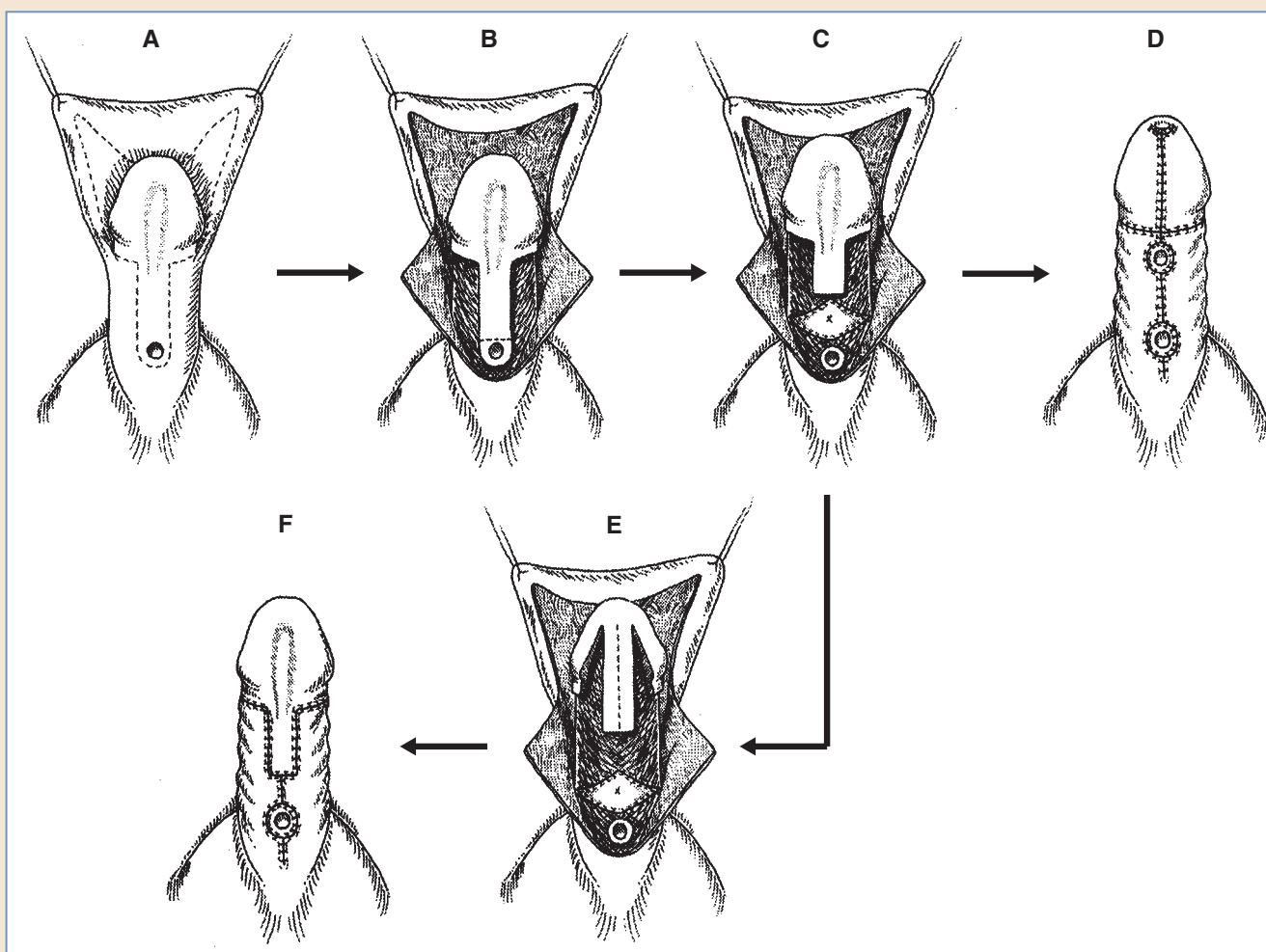


Fig. 61-11 Proximal division of the urethral plate. **A**, An incision is made along the inner preputial skin to fashion a mucosal collar, and a U incision is made along the penile shaft, preserving the urethral plate. The base of the U incision is behind the hypospadiac meatus. **B**, After the chordee is corrected, if division of the urethral plate and staged repair will be necessary, the plate is divided proximally at the level of the original hypospadiac meatus. **C**, Chordee tissue, fibrous tissue along the ventral penis, is further dissected and resected. This usually results in distal migration of the urethral plate and proximal migration of the hypospadiac meatus. Correction of chordee is completed with dorsal plication and/or corporal body grafting (a diamond-shaped graft) at the point of maximal curvature. **D**, The urethral plate is undisturbed, and Byars flaps are used to fill the gap between the plate and hypospadiac meatus. The plate is tubularized at the second stage. **E**, Alternatively, the urethral plate is incised and tubularized using the Snodgrass technique at the first stage repair. **F**, Byars flaps are used to cover the defect between the hypospadiac meatus and the tubularized urethral plate.

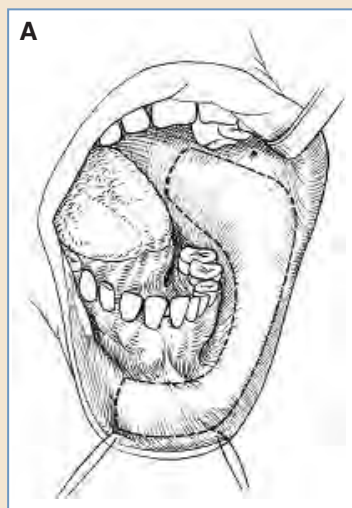
Reoperative Hypospadias

When primary hypospadias repairs (single-stage or staged) fail or break down, subsequent attempts at repair can be very difficult. When the urethral plate is still intact or the surrounding tissue appears viable and healthy, a repeat repair with one of the techniques described previously can be successful. However, in patients who have undergone numerous failed attempts at repair, the anatomy and native penile tissue are often distorted and the blood supply compromised. Predicting the tissue reaction with subsequent surgery is challenging. These patients are known as *hypospadias cripples* and usually have a severe deformity of the penis.⁵¹ These are among the most difficult and technically challenging cases for hypospadias surgeons. Repairs must be individualized based on the tissue available for use and the functional needs and desires of the patient.

For all reoperative hypospadias patients, the degree of residual chordee is corrected first. Next, tissue available for creating a new urethra is assessed. Well-vascularized penile skin that can be used is usually the first choice for a urethral substitute. When the urethral plate and/or local penile skin is not suitable because of fibrosis and scarring, an alternative source of tissue is needed. Historically, surgeons have employed numerous free grafts with variable success, including non-hair-bearing skin and bladder mucosa.^{52,53} Most recently, buccal mucosa was found to be the most successful tissue for use as a free graft.⁵⁴⁻⁵⁶ As a urethral wall replacement, it has superior properties that promote neovascularization and histologic features that most closely resemble native urethral tissue, compared with other free grafts.⁵⁵ Harvesting buccal mucosa from the inner cheek and/or lower lip is a relatively simple procedure with minimal morbidity. Even large grafts can be harvested without closure of the defect within the oral mucosa, because the oral mucosa regenerates quickly and rarely leaves a constrictive defect.⁵⁷ In patients with a viable urethral plate, a buccal mucosa graft can be placed on the plate in an onlay fashion as a single-stage procedure. Because this is a free graft and requires neovascularization, coverage with well-vascularized tissue such as a tunica vaginalis flap is essential for success^{58,58a} (Fig. 61-12).

In cases in which no healthy urethral plate is available, a two-stage buccal mucosa graft urethroplasty is employed. At the first stage, the unhealthy urethral plate and surrounding scar tissue are excised, and the buccal graft is secured and quilted to the bare penile tissue along the

Fig. 61-12 Buccal graft urethroplasty.
A, A graft of buccal mucosa is harvested from the inner portion of the cheek or the lower lip. Care is taken to prevent injury to Stensen's duct, which is adjacent to the second molar.



Continued

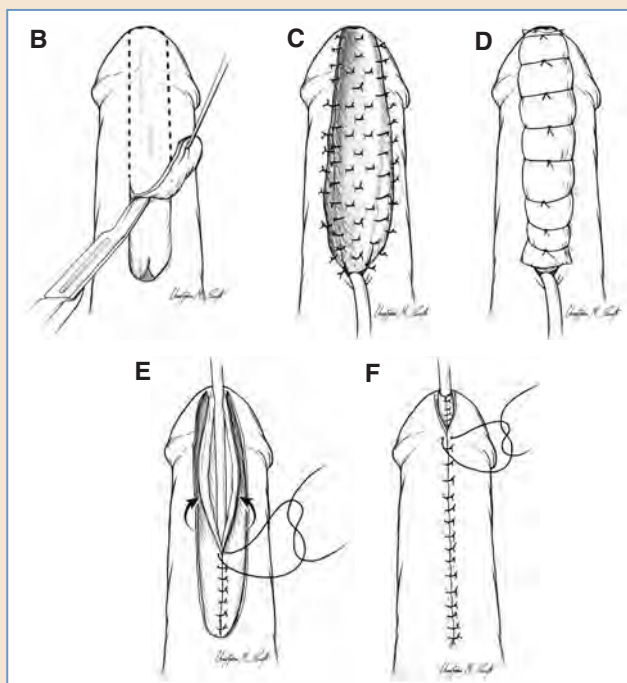


Fig. 61-12, cont'd **B**, Scarred unhealthy tissue is removed. The glans is split widely and the shaft skin elevated laterally to facilitate a wider well-vascularized bed for grafting. **C**, The defatted buccal graft is quilted into place from the proximal urethral meatus to the tip of the glans and the elevated lateral skin with as wide a graft as possible. **D**, A bolster is secured to the ventral aspect of the penis to improve graft take during revascularization. **E**, After 6 months the buccal mucosa is tubularized over a stent, with either dartos or tunica vaginalis flaps covering the suture line. **F**, The ventral aspect of the penis is resurfaced with Byars flaps.

ventral surface of the penis. The graft is fenestrated to decrease the risk of hematoma formation and to enhance imbibition of the graft. Six months later, after the graft develops adequate blood supply from the surrounding tissue, it is tubularized (see Fig. 61-12). Two-stage buccal mucosa urethroplasties have been shown to have greater success and superior cosmesis, compared with historical repairs for reoperative hypospadias. Long-term evaluations has shown a complication rate of 24% to 37% with no recurrent ventral curvature even in patients followed longitudinally after puberty.^{57,59,60}

POSTOPERATIVE CARE

At the completion of hypospadias repairs, most surgeons place a postoperative dressing for the phallus. The type of dressing is based on the surgeon's preference. An ideal dressing is nonadherent, provides slight compression to prevent swelling and/or to promote hemostasis, and absorbs wound drainage. At our institution, the surgical site is wrapped with a dressing of benzoin-soaked Owens gauze and then Coban secured with clear plastic meshed tape. This dressing provides gentle compression and keeps the wound clean and dry. It is left in place for 4 to 5 days and removed

in the office by a physician or nurse. Others have advocated a transparent bioocclusive dressing such as Tegaderm, silicone foam dressings, and recently, octyl cyanoacrylate (superglue).⁶¹⁻⁶³ Two studies have compared different types of dressings to no dressing after hypospadias repair.^{64,65} Both found that complications did not increase postoperatively when only antibiotic ointment (and no other dressing) was used; they concluded that a dressing was not required after routine hypospadias repairs. In contrast, some procedures are more extensive and require a great deal of dissection; therefore postoperative hemostasis is an issue. In these cases a more involved elastic dressing that places pressure on both the penile repair and the perineum may be extremely valuable.⁶⁶ Once a dressing is removed, a topical antibiotic ointment is typically applied to the penis for 1 or 2 weeks to create a barrier between the wound and the diaper and its contents while the suture lines heal.

Temporary urinary diversion with a urethral stent (6 to 8 Fr) is commonly used after hypospadias repair to direct urine to drip into the child's diaper. Stents are usually left in place for 7 to 14 days postoperatively. Types of urethral stents include a pediatric feeding tube and commercially available tubes such as the Firlit-Kluge urethral stent, the Zaontz urethral stent, and the Koyle diaper stent, all from Cook Medical. Infection is a concern with open-ended stents; however, Montagnino et al⁶⁷ found no differences in the rates of postoperative urinary tract infection when open urethral stents with double diaper techniques were used, compared with a traditional closed urinary drainage system. Stents are thought to facilitate adequate healing of the neourethra. Whether stents are needed in routine cases of hypospadias is not clear. Several studies have suggested that the complication rate is not increased when a stent is not used.^{68,69} Not only does stenting decrease postoperative urinary retention, but also some studies suggest that stenting promotes a higher success rate. It decreases postoperative urinary retention. Buson et al⁷⁰ found that complications after Mathieu repair were significantly higher (18.9% versus 4.6%) in unstented versus stented repairs, and El-Sherbiny⁷¹ reported a higher reoperation rate in patients with unstented Snodgrass repairs than in patients with stented repairs. Some surgeons advocate suprapubic urinary diversion. One randomized trial compared the use of a suprapubic catheter and a small anterior urethral catheter versus urethral urinary drainage and noted decreased bladder spasm (33% versus 0%), a lower rate of fistula (12.7% versus 2.3%), and similar meatal stenosis rates (2.4%), respectively.⁷²

Most pediatric urologists prescribe antibiotics after both unstented (60.2%) and stented (76.6%) hypospadias repairs.⁷³ This practice has been questioned because of increased focus on antimicrobial stewardship and the conflicting results of studies evaluating the benefits of routine antibiotic prophylaxis after stented hypospadias repairs.⁷³⁻⁷⁵ In a practice environment where judicious antibiotic use is required to prevent resistant microorganisms, a prospective multicenter randomized controlled trial currently enrolling patients at our institution will potentially clarify the role of antimicrobial prophylaxis after stented hypospadias.

TREATMENT OUTCOMES AND COMPLICATIONS

The overall outcome and complication rate after hypospadias repair is generally directly proportional to the length of urethra reconstructed and the overall complexity of the repair. Possible complications after hypospadias repair include bleeding or hematoma formation, infection, urethral fistula, meatal stenosis, recurrent chordee, urethral diverticulum, partial or complete urethral repair breakdown, that is, dehiscence, urethral stricture, and erectile dysfunction. The most common complications for distal hypospadias repairs include fistula (3.8% to 11.5%), meatal stenosis (0.7% to 7.7%), wound dehiscence (1.6% to 2.6%), and urethral stricture (0% to 0.7%).^{30,76} Proximal repairs have higher rates of complications, including fistula (15% to 33%), meatal stenosis (26%), chordee (18%), dehiscence (11.3% to 42.3%), urethral diverticulum (4%

to 27%), and obstructive voiding symptoms (37%).^{77,78} In general, unless immediate intervention is required to address postoperative bleeding, infection, or wound debridement, reoperation for hypospadias should be delayed for at least 6 months to allow edema and inflammation to subside and full healing to occur.

The most common complication after hypospadias repair is urethrocutaneous fistula formation. Overall fistula rates range from 3.8% to 33%.^{76,77} As mentioned previously, the more proximal the hypospadias repair, the higher the risk of fistula formation. Recent advancements in surgical technique, optical magnification, and suture material have significantly lowered the fistula rate. It is relatively uncommon for a fistula to develop in an uncomplicated repair of distal hypospadias. A variety of factors may be responsible for fistula formation, including infection, tissue ischemia, and meatal stenosis. Fistulas are most frequently apparent within the first year after the repair. However, up to 23% become evident only after 1 year of follow-up.^{79,80} They generally manifest as urine leakage from an area of inflammation or wound breakdown on the ventral surface of the penile shaft. Rarely, a small fistula will close spontaneously with observation and local wound care, but most often a formal surgical repair of the fistula is required once it fully epithelializes. Fistula repair is best performed 6 to 12 months after the initial repair to allow resolution of tissue edema and induration. Small fistulas can usually be repaired by excision of the fistulous tract down to healthy tissue, inversion of fresh edges, primary closure, and coverage with multiple layers of well-vascularized surrounding tissue. In cases in which a simple primary closure is performed, distal stenosis/stricture should be ruled out as a cause of the fistula. When this is present, a complete repeat of the hypospadias repair may be needed. Larger fistulas may require more extensive repair with the use of onlay flaps, revision of the glans closure, and/or a possible complete repeat of the primary repair if the remaining intact neourethra is stenotic or poorly formed.

Meatal stenosis that develops after a hypospadias repair is probably related to technical aspects of the neomeatus creation, for example, tension on the neomeatus is excessive, or the meatus is too narrow. Snodgrass⁸¹ described the following methods for preventing this complication in a TIP procedure: not tubularizing the neourethra too far distally into the glans, making sure that the incision of the urethral plate is deepened down near the level of the corporal bodies, and resisting the urge to extend the relaxing incision into the distal glans and the area of the neomeatus. When these recommendations are followed, meatal stenosis is relatively uncommon. In a large series of TIP repairs, the meatal stenosis rate was 1%.³² Mild forms of this complication can be treated by the parents with a short period of frequent dilation using the tip of an ophthalmic ointment tube or a small-caliber feeding tube.⁸² A topical steroid ointment can be used in conjunction with dilation to decrease edema and fibrosis. More severe or recurrent forms may require a formal meatoplasty.

A urethral diverticulum can form postoperatively as a result of distal obstruction, meatal stenosis, general laxity of the neourethral tube from redundant tissue, and/or skin that stretches more than expected. This problem rarely occurs in repairs that primarily use the urethral plate, and it is more common with long neourethras made from local skin flaps. Symptoms from diverticulum formation include ballooning of the urethral tube while voiding, dribbling of retained urine after voiding, and urethral stone formation. In patients with distal obstruction, correction

of the outflow obstruction may improve these symptoms and resolve the diverticulum when it is discovered early. However, excision and repair of the urethra is usually necessary once a diverticulum has formed.

Urethral strictures are typically found within the first year after hypospadias surgery and manifest most frequently in the area of the original hypospadiac meatus.⁸⁰ Many factors can contribute to stricture formation, including tension on the urethral tube, tissue ischemia, inappropriate formation of the neourethra, infection, trauma from instrumentation, and the use of large catheters.⁸² Symptoms include urinary hesitancy, decreased caliber of the urinary stream, straining to void, and/or infection. A diagnosis is usually made with retrograde urethrography and/or cystoscopy. Mild strictures can be managed with intermittent urethral dilation when diagnosed early. An internal urethrotomy may be helpful for short strictures without dense scar formation. Generally, most strictures will not respond to dilation or incision unless they are very mild. For strictures that recur after one dilation or incision, either excision of the stricture or a formal repeat urethroplasty is indicated.

Previously, surgeons evaluated the success of hypospadias repairs, based almost entirely on their own cosmetic and functional outcome criteria. The use of patient- and parent-reported quality-of-life outcomes and satisfaction measures is increasing. These data provide objective measures for the outcomes that mean the most to patients. Wide adoption of objective scores, such as the Hypospadias Objective Penile Evaluation score and the Penile Perception Score, will improve reporting of surgical outcomes.^{83,84} Because mild forms of hypospadias do not necessarily “need” to be repaired, these outcomes are especially important for counseling. As expected, currently reported quality-of-life outcomes are worse with more proximal/severe hypospadias. Surveys have shown decreased satisfaction with cosmetic appearance, decreased satisfaction with penile length, worse erectile dysfunction (distal: 9%, midshaft: 50%, and proximal: 72%), and impaired sexual quality of life in patients with proximal hypospadias.^{85,86}

FUTURE DIAGNOSTIC, ASSESSMENT, AND TREATMENT MODALITIES

Hypospadias is a surgical disease. In the future, the current goals of repair will endure: correcting the chordee, repositioning the urethral meatus in an orthotopic location, and creating an overall excellent cosmetic appearance of the penis. Corrective surgery for hypospadias has evolved. Recent advances include maximizing the use of a patient’s native urethral plate, an increased awareness of the importance of a second layer of coverage over the repair, and a better understanding of the abnormal anatomy. These changes have greatly improved the overall success rates, decreased complications, and enhanced the functional and cosmetic outcomes. Despite recent improvements in surgical technique, cases of proximal hypospadias continue to be a technical challenge for even the most experienced surgeons. Hopefully, further technical and surgical advancements will result in success rates more in line with current success rates for distal hypospadias. New discoveries in tissue engineering research and basic science work focusing on cellular factors important for optimal wound healing hold great promise for improving our current surgical armamentarium for hypospadias repair.

KEY POINTS

- Many types of hypospadias repair have been developed over the last several decades with varying degrees of success.
- Currently, the most commonly used repairs that lead to excellent results emphasize functional success and cosmesis.
- A successful repair should result in a straight penis with a urethral meatus properly positioned at the tip of the glans and a normal-appearing penis.
- Most routine cases of hypospadias can be performed on an outpatient basis in patients who are at least 6 months of age, with a success rate greater than 95%.
- Surgical success depends on a tension-free repair using well-vascularized tissue with additional vascularized flap coverage.
- For more severe forms of hypospadias, a two-stage repair may be needed.
- The decision to perform a one-stage or two-stage repair depends on the degree of chordee and the integrity of the urethral plate.
- In patients without an adequate urethral plate or penile skin to create a neo-urethra, including those requiring reoperative hypospadias, repair with buccal mucosa has been very successful.

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62

Feminizing Genital Surgery in Disorders of Sex Development

Elizabeth B. Yerkes



Feminizing genital surgery for intersex conditions, or disorders of sex development in current consensus terminology,¹ has evolved tremendously over the past half century.^{2,3} As emphasis was placed on functional potential and feminizing cosmetic outcomes, technical modifications based on the gradually growing understanding of clitoral neurovascular anatomy and its likely relationship to sexual function led to the nerve-sparing techniques most commonly used today. In the middle of the last century, clitoral reduction was effectively *clitorectomy*. This was recognized as unacceptable because of the resulting difficulties with arousal. The next iteration of feminization was *clitoral recession*, with the clitoral bodies folded on themselves for concealment. Although this procedure feminized and preserved the clitoral tissues, arousal and engorgement resulted in a painful bulge in the infra-pubic and mons regions. Subsequently, over the past several decades, a more detailed understanding of the clitoral neurovascular anatomy has led to a series of modified techniques collectively called *reduction clitoroplasty*. These more contemporary techniques attempt to protect the integrity and innervation of the glans clitoris and the proximal neurovascular supply to the clitoris with the goal of preserving sexual function.

This chapter discusses the following components of feminizing genitoplasty: degloving, management of the urethral plate, management of the clitoral body, recession of the glans and creation of the clitoral hood, and labioplasty. Depending on whether vaginoplasty is undertaken in the same setting as feminizing genitoplasty or deferred, as with congenital adrenal hyperplasia, modifying the appropriation of tissue resources and minimizing future use of previously operated tissues must be considered.

PREOPERATIVE CONSIDERATIONS

The surgical approach and considerations are also applicable to male-to-female transgender feminization in adults, with the caveat that the tissues for creation of the clitoral hood and labioplasty will likely be affected by chronicity of androgen exposure and resultant tissue changes and rugation despite preoperative conversion to an estrogen environment. Nerve-sparing techniques with glans reduction and penile skin-flap vaginoplasty have been reported with good results.⁴ Some of these tissue changes may also be seen in older children and adolescents with poor medical control of congenital adrenal hyperplasia or disorders of sex development, in whom surgery is deferred until the patient can participate in the decision-making process.

The timing and indications for clitoroplasty and vaginoplasty in infants born with disorders of sex development are currently quite controversial. The 2006 “Consensus statement on management of intersex disorders”¹ suggested “surgery should only be considered in cases of severe virilization (Prader III, IV, and V) and should be carried out in conjunction, when appropriate, with repair of the common urogenital sinus.” Surgeons performing these reconstructive procedures must be well versed in the physiology of these conditions and the current controversies, practice patterns, and potential legal implications with regard to irreversible genital surgery in infants and minor children. A strong understanding of clitoral anatomy is also essential. This chapter is written as a guide for surgical concepts of feminizing genitoplasty for external structures, if reconstruction is elected after extensive counseling regarding the known (cosmetic, secondary procedures) and potentially unknown (sexual function and gender dysphoria) outcomes. Informed decision-making for surgical intervention or actively deferred surgical intervention is a highly individualized process.⁵ This chapter is written neither in support of nor in opposition to genital reconstructive procedures. As stated previously, reconstruction of the urogenital sinus to externalize the urethra and vagina as separate openings is often undertaken at the same time as the external aspects of feminizing genitoplasty. The timing is controversial primarily because of concerns regarding the need for secondary procedures, namely, secondary procedures for vaginal stenosis. If the parents do not elect external feminizing surgery during their child’s infancy, reconstruction of the urogenital sinus is also often deferred. The fine points of the various approaches to the urogenital sinus and forms of replacement vaginoplasty are beyond the scope of this chapter on external elements of feminizing procedures. Some details will be provided as relevant to surgical planning for feminizing genitoplasty.

CLITORAL ANATOMY AND NEUROANATOMY

Our current knowledge of clitoral neuroanatomy is derived from the work of Baskin and colleagues^{6,7} on sections of the human fetal clitoris, as well as their work with a hyena model of clitoral hypertrophy. The cavernous (autonomic) nerves are responsible for erectile function of the clitoris. Sensation of the clitoris is from the paired dorsal nerves of the clitoris. The cavernous nerves originate from the vaginal plexus (related to the inferior hypogastric plexus of the rectum) at the 10 o’clock and 2 o’clock positions along the anterior vagina and course along the 5 o’clock and 7 o’clock positions of the proximal urethra. The cavernous arteries travel with the nerves. The cavernous nerve and artery enter each clitoral body under the pubic arch near the union of the crura to form the clitoral body. Branches of the cavernous nerve also join the dorsal nerves under the pubic arch.⁸

The dorsal nerves originate from the pudendal nerves (somatic sensory and motor) and course along the crura and ischiopubic rami. Under the pubis at the bifurcation to the crura, the dorsal nerve maintains a lateral position. The relationship between the crura and dorsal nerve is different from the neuroanatomy in the male. Dissection between the crura and rami should be avoided in females.⁸ Dorsal nerve tissue is minimal in the midline, but nerve tissue is more notable at the

11 o'clock and 1 o'clock positions along the clitoral body. The dorsal nerve branches fan dorsolaterally but are deficient ventrally^{6,7} (Fig. 62-1). The glans clitoris has greatest innervation dorsally but minimal ventrally.⁶ In a virilized clitoris, as in patients with congenital adrenal hyperplasia, variations from normal may be expected. The hyena model suggests that the nerves may indeed course more laterally; this supports the use of ventral incisions at the stage of reduction of the clitoral body (see the section Management of the Clitoral Body).⁷

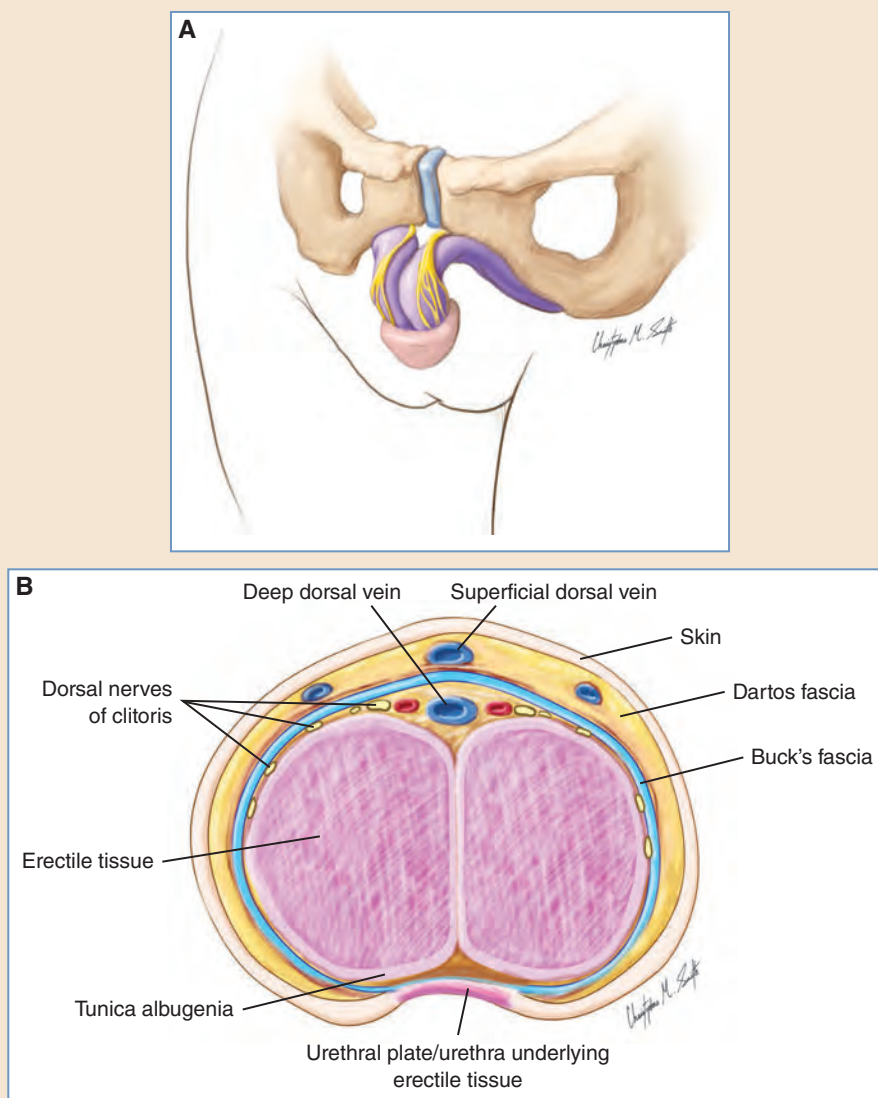


Fig. 62-1 **A**, A dorsal view of a hypertrophied clitoris in a patient with a disorder of sex development. The relationship between the nerves and pubic arch proximally is shown. The dorsal midline has no nerves, but they are concentrated dorsolaterally on each side. The nerves fan laterally. **B**, A cross-section of the clitoris. The bulk of the erectile bodies changes with the degree of virilization. The layers most pertinent to the degloving step are the dartos and Buck's fascia. In this illustration and the subsequent cross-sections, the dartos is depicted as distinct from the subcutaneous tissue, but they are effectively a single layer and envelop the superficial veins. Immediately deep to the Buck's fascia, the dorsal vein and arteries are closer to the midline of the body of the clitoris, and the nerves are more dense dorsally, present but less dense laterally, and deficient ventrally.

O'Connell et al⁹ provided important anatomic information about the true clitoral anatomy. Specifically, MRI studies in nulliparous premenopausal women revealed the natural hairpin curve of the clitoris, which results in a more caudal position of the glans relative to the clitoral body. The erectile components of the clitoral complex were clearly identified, including their relationship to the pubic arch and the urethra and vagina. The erectile complex includes paired clitoral bodies with a thin, fibrous septum distally and separation proximally to form the paired crura and paired bulbs, which nearly join ventral to the urethra and flank the urethra and vagina distally. Previous depictions of the female anatomy showed this as less defined tissue⁹ (Fig. 62-2).

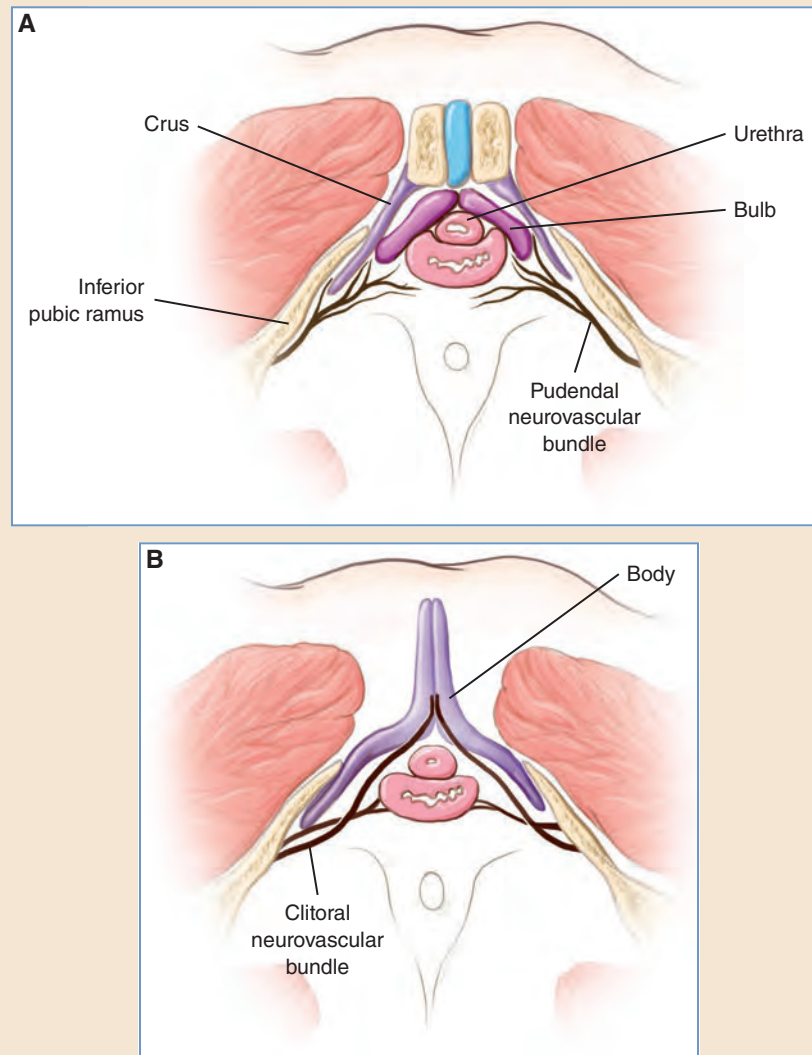


Fig. 62-2 Normal pelvic and clitoral anatomy. **A**, An axial view shows the relationship between the clitoral erectile cluster (body, crura, and bulb) and the urethra. Pudendal neurovascular tissue is also shown. **B**, The clitoral (cavernous) nerves enter the clitoral body medially near the bifurcation.

COMPONENTS OF REDUCTION CLITOROPLASTY

General Concepts

Feminizing surgery deserves the same meticulous attention as masculinizing reconstructive procedures. Feminization is equally delicate despite its more deconstructive nature. Surgeons should understand the clitoral neurovascular anatomy and the wide spectrum of “normal” female genital anatomy before performing feminizing genitoplasty. Balancing cosmesis and functional outcomes is important; surgeons must keep in mind that resources are highly variable and may place limits on achieving a normal appearance. Optimizing the potential for intact sexual function is essential, but the patient or parents by proxy must understand that sexual function may be impaired because of nonanatomic or nonphysiologic factors. Multiple discussions regarding goals and reasonably expected short-term and long-term outcomes are critical. Sharing of medical information and psychological support are key elements of the work of surgeons and multidisciplinary teams.

Degloving

All adhesions between the glans clitoris and inner prepuce are bluntly lysed. A traction suture of 4-0 or 5-0 Ethibond is placed in the dorsal midline of the glans (Fig. 62-3). Early and late steps are taken to adequately conceal the clitoris and to fashion a cosmetically appropriate clitoral hood. The first of these steps is to choose a site for the degloving incision. Unlike penile procedures such as circumcision or hypospadias repair, in which the incision may be 6 to 8 mm from the glans margin, the initial incision for clitoroplasty should allow sufficient inner preputial tissue to cover the glans (Fig. 62-4). Although surgeons should recognize that the inner preputial tissue has properties suitable for the vestibular aspect of the labia minora, this tissue should be shared to create a clitoral hood that minimizes exposure while preventing entrapment of the glans. The outer prepuce and skin of the hypertrophied clitoris will ultimately be used to fashion the labia minora (see the section Labioplasty). The clitoral skin and clitoris itself are the most sensitive tissues in adults with normal clitoral anatomy¹⁰ and therefore should be fully preserved in reconstructive procedures.

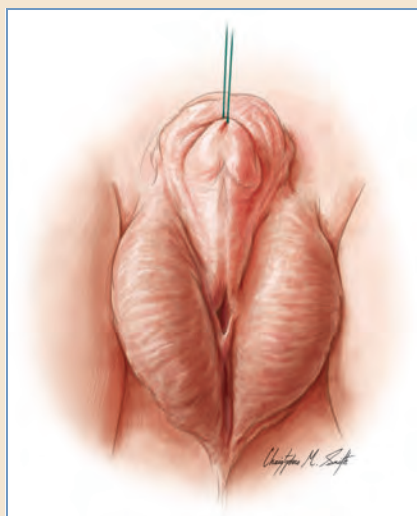


Fig. 62-3 The traction suture dorsally allows exposure of all elements of dissection. Ventral curvature still results in some rotation of the clitoral body early in the dissection; additional manual retraction may be helpful for ventral dissection. The degree of virilization is variable; in this case, the fusion and rugation of the labia majora and location of the urogenital sinus proximal to the base of the hypertrophied clitoris are shown.

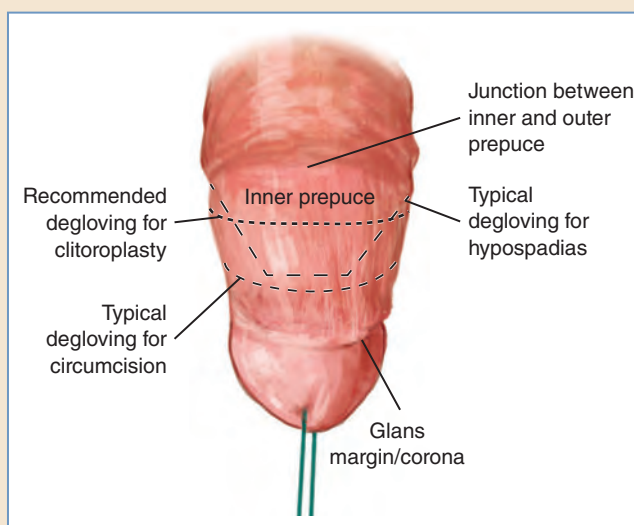


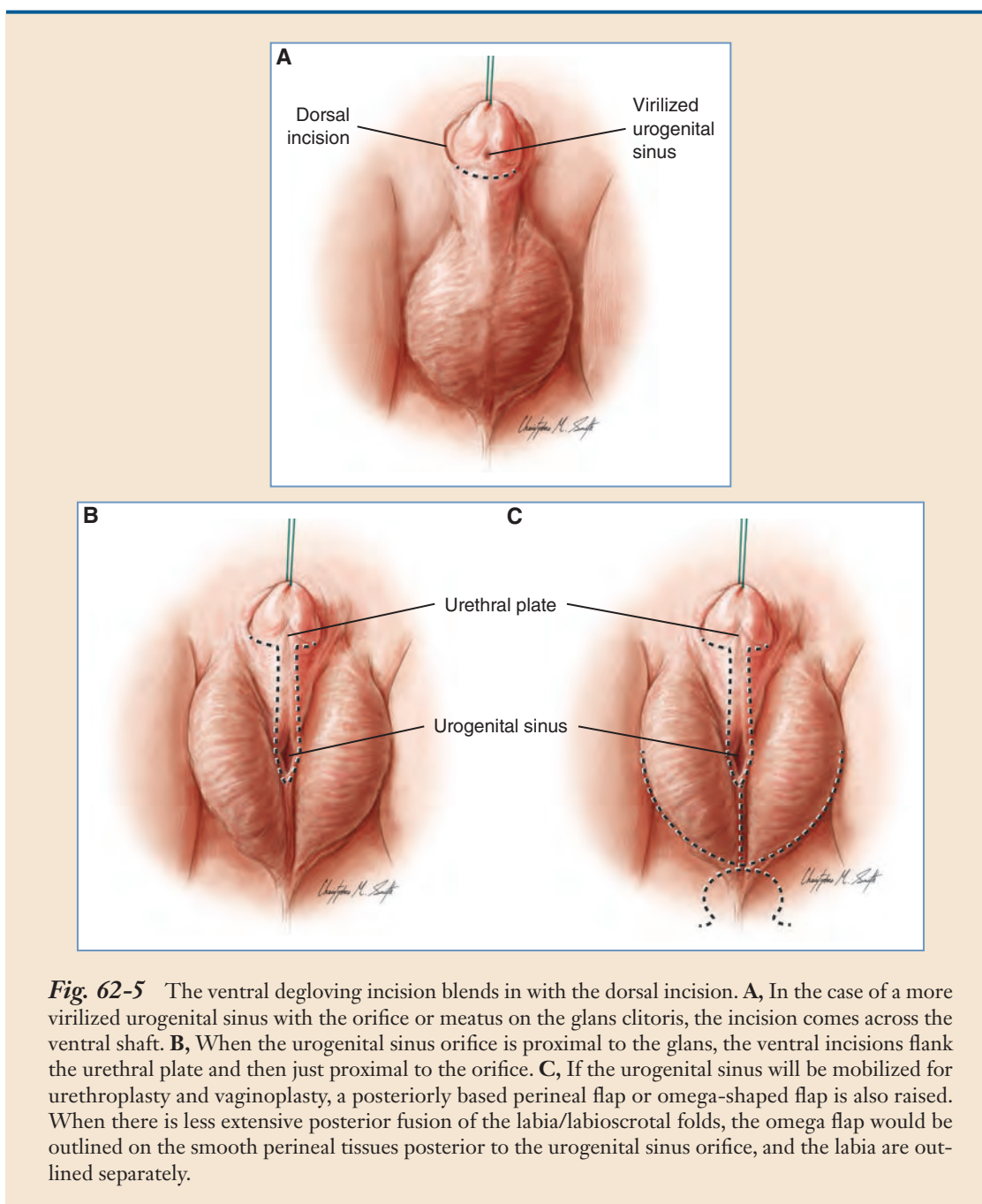
Fig. 62-4 The traction suture allows exposure dorsally for degloving. The clitoral preputial adhesions have been lysed, and the skin is retracted to expose the inner prepuce. The difference in character between the inner and outer prepuce is shown. The location of the degloving incision for the clitoroplasty as opposed to the standard incisions for circumcision or hypospadias in a male child is shown. The dorsal incision for clitoroplasty should allow adequate skin to cover the glans; this is the first step in constructing a clitoral hood.

The ventral incision comes across the shaft distally if the urethra is virilized with a glanular or coronal meatus, or it flanks the urethral plate in less virilized patients¹¹ (Fig. 62-5, *A* through *C*). If mobilization of the urogenital sinus for urethroplasty and vaginoplasty is also undertaken, the incision extends proximal to the urogenital sinus orifice. A posteriorly based perineal flap or omega-shaped flap is created to assist in dissection of the urogenital sinus. This flap is also reserved for incorporation into the spatulated posterior wall of the vagina to create a wider vaginal orifice and to reduce the risk of cicatrix formation. Certain approaches to vaginoplasty may not use this tissue, or the unique anatomy may not require the use of this flap. The pros of having this tissue available have been stated previously, but the drawback is that this tissue may be hair-bearing in the future and does not have mucosal properties typical of the vagina. Incisions are also created around the base of each labia majora to judiciously reduce the labia and to move them posteriorly to flank the new vaginal orifice.

The clitoris is sharply degloved to the base, dissecting the dartos with the skin, but leaving Buck's fascia and its underlying neurovascular tissue undisturbed. The clitoral length is often markedly increased by degloving and division of the urethral plate. The dorsal suspensory ligament should be left intact to support the glans in a natural position after reduction is complete.

Management of the Urethral Plate

After degloving the clitoris, the urethral plate with its underlying spongiosal tissue will be mobilized in preparation for work on the clitoral bodies. If the urethra is intact, it must be divided on the distal shaft and mobilized off the clitoral body before clitoral reduction.



Decisions will be made at this point based on the unique anatomy and the total surgical plan. If a urogenital sinus is present and a single-stage repair is elected, urethroplasty and vaginoplasty will be performed, along with feminizing genitoplasty. The urogenital sinus work will be performed after the initial clitoral preparation. The urethral plate will be divided distally and dissected off the clitoral body. Where it is divided may depend on the projected appropriation of the urethral plate and urogenital sinus tissue. Will the mobilized urogenital sinus be used for a portion of the vaginal wall, or will the sinus tissue be available for a mucosa-lined vestibule?^{11,12}

Will mobilization of the sinus for urethroplasty and vaginoplasty be deferred? In this case, the urethral plate is divided and partially resected to allow recession of the glans, while an adequate mucosal strip is maintained between the glans and the urethra⁶ (Fig. 62-6).

In patients with significant chordee and a short urethral plate in whom urogenital sinus mobilization will not be undertaken, the plate can be elevated and preserved without division. After the clitoral body tissues are reduced, the plate is returned to its natural location⁶ (Fig. 62-7). In addition to the natural mucosal strip separating the clitoris from the urethra, the intact plate may allow one additional source of perfusion for the glans.⁶

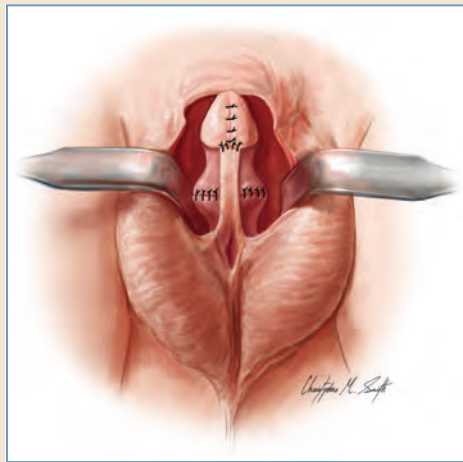
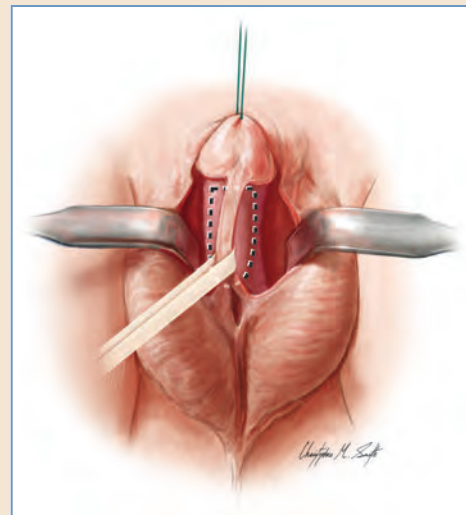


Fig. 62-6 The urethral plate is divided distally and then preserved in the child in whom clitoral reduction is performed but repair of the urogenital sinus is deferred. Note that the glans was reduced ventrally in this figure.

Fig. 62-7 When mobilization of the urogenital sinus will be deferred or is not required (based on a patient's unique anatomy and the surgical plan), the urethral plate may be elevated for clitoral body reduction without actually dividing the plate. The plate can be returned to its natural position without generating bulk or a bulge in this region only in patients with prominent chordee and a short plate.



Management of the Clitoral Body

Nerve-Sparing Clitoral Reduction Techniques

Several different nerve-sparing techniques are currently used, based on surgical training and preference. These approaches support the glans on an intact dorsal neurovascular pedicle while allowing debulking of the hypertrophied clitoral body.

In one approach, Buck's fascia and the neurovascular tissue are elevated as a unit off the dorsal aspect of the tunica albuginea. The glans remains attached to the neurovascular tissue but is dismembered from the two corporal bodies. Poppas et al¹³ have called this approach *nerve-sparing ventral clitoroplasty*. Buck's fascia is incised ventrally to minimize the risk of injury to the dorso-laterally fanning nerves. Dissection is carried out between the tunica albuginea and the neurovascular tissue circumferentially along the length of the clitoral body (Fig. 62-8, A).¹³ Papaverine is used to minimize spasm of the delicate vasculature. The erectile bodies are dissected, excised to within 1.5 to 2 cm of the bifurcation, and oversewn (Fig. 62-8, B).¹³ The glans is then seated onto the proximal corporal stump. The stump allows proximal engorgement and support of the glans. A vascular anastomosis is not performed. Great care is required to prevent kinking of the neurovascular tissue as the glans is reseeded.

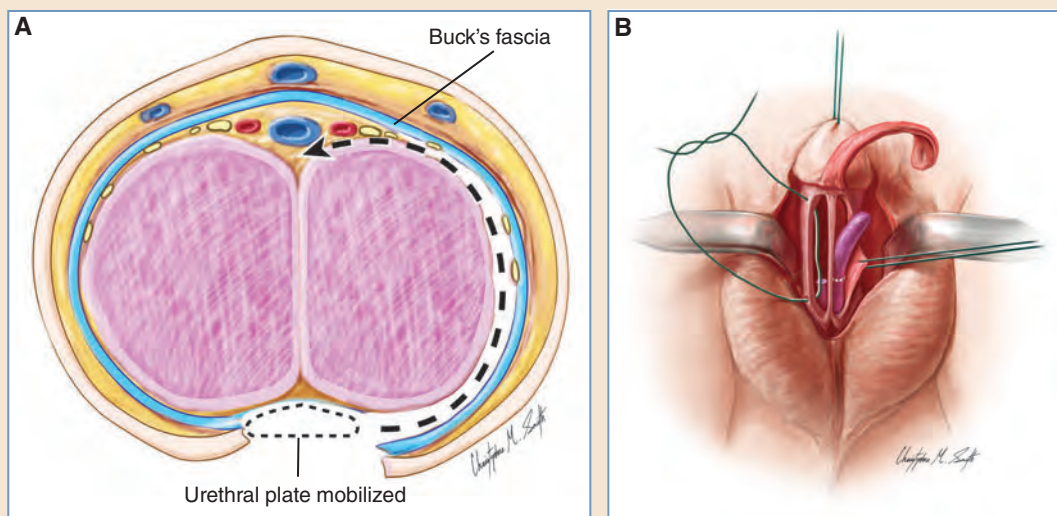


Fig. 62-8 Nerve-sparing ventral clitoroplasty. **A**, A cross-section of the clitoris. Buck's fascia is incised ventrally, and the dissection occurs between the tunica albuginea and the dorsal neurovascular tissues. Circumferential dissection along the clitoral body results in a sheath of neurovascular tissue and Buck's fascia. **B**, Glans and nerves have been elevated off the erectile bodies. Bodies are transected and excised proximally. The glans will be reseeded on the oversewn proximal tissue. The surgeon must avoid kinking of the neurovascular tissue.

Another often-endorsed nerve-sparing approach leaves the dorsal and lateral tunica albuginea intact; meanwhile, the neurovascular tissue maintains its natural position between Buck's fascia and the tunica albuginea (Fig. 62-9, *A*). Longitudinal incisions are created bilaterally through the tunica albuginea ventrally. The erectile tissue is dissected as a unit from inside the rind of the tunica albuginea with sharp and blunt dissection.⁶ The septum may be traversed and resected with the spongy erectile tissue of both sides, or each side may be dissected separately. The tissue is dissected from the distal tips of the clitoral bodies down to within 1 cm of the bifurcation. The spongy cavernosal tissue is ligated with absorbable suture and excised (Fig. 62-9, *B*),⁶ and the glans is seated on the corporal stump. I prefer this approach over nerve-sparing ventral clitoroplasty,¹³ because the risk of inadvertent neurovascular injury, despite meticulous technique, is theoretically reduced and the residual tissue is not notably bulky.

Because of the known neurovascular anatomy,⁸ dissection between the erectile bodies at the bifurcation or encircling the crura at the bifurcation to fashion a tourniquet is not recommended.

One additional contemporary option exists. This approach preserves all clitoral tissue until the child has reached the age of consent or well-informed assent and can choose formal reduction or even reconstruction to whole, as desired. Pippi Salle et al¹⁴ described a novel clitoroplasty technique in which the glans clitoris and dorsal neurovascular tissue are dissected as a unit off the clitoral body. The clitoral body is then split down the midline, and each erectile body is rotated and buried in its ipsilateral labium majus as the labial work is completed. It is not yet known whether engorgement of the repositioned but retained clitoral bodies will be pleasurable, painful, both, or neither.

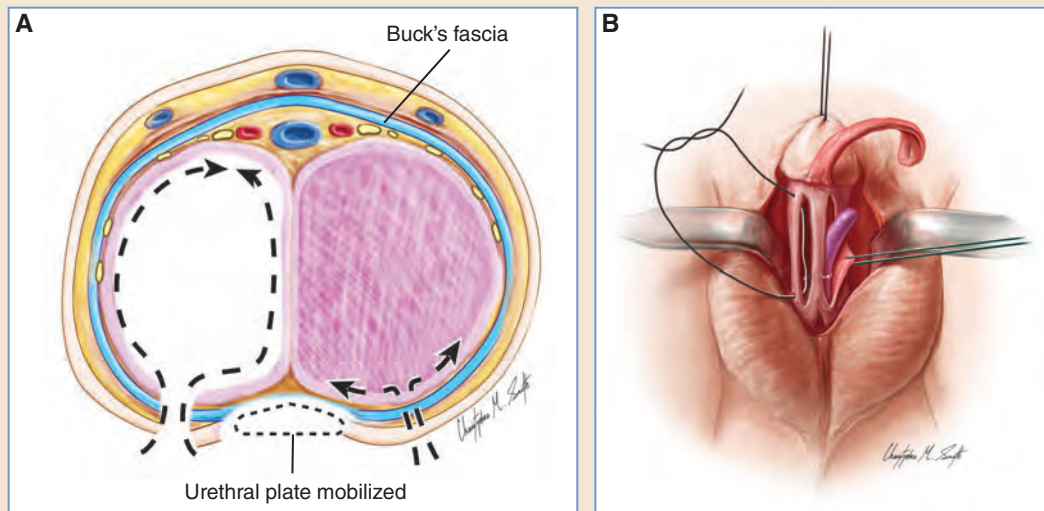


Fig. 62-9 Another common nerve-sparing approach for clitoral reduction. **A** and **B**, A ventral incision prevents injury to the nerves. An incision through the tunica albuginea facilitates removal of the spongy erectile tissue without resecting the tunica albuginea or dissecting near the neurovascular tissue.

Recession of the Glans and Creation of the Clitoral Hood

Reduction of girth or diameter of the glans is largely the surgeon's preference. In pediatric feminizing genitoplasty, I typically do not reduce the glans, because there is no clear evidence that a well-concealed but larger than typical glans is medically harmful or otherwise bothersome. Great caution has just been taken to preserve viability and sensation, and reduction of the glans *could* have an adverse impact, even if the surrounding tissue maintains sensation. Achieving a glans clitoris of entirely normal proportions is not feasible in patients having clitoral reduction. Focus is redirected from size to recession of the glans and concealment with a reconstructed hood and labia to achieve a feminine cosmetic result that minimizes overstimulation of the sensate, reconstructed glans clitoris. If reduction is necessary in a very large glans, a ventral wedge resection is least likely to affect nerves.

A clitoral hood is fashioned in two stages as the glans is recessed. The inner prepuce preserved at the time of the initial skin incision is brought up and over the glans and configured to a prepuce-lined hoodie shape (Fig. 62-10, *A* and *B*).

The glans is positioned next. If the dorsal tunica albuginea has been left intact, the glans is recessed and secured to the proximal clitoral bodies by suturing together the proximal and distal margins of the corporotomies. This is done more to provide support than hemostasis. The neurovascular tissue positions itself in the subcutaneous space at the dorsum of the clitoris, near the

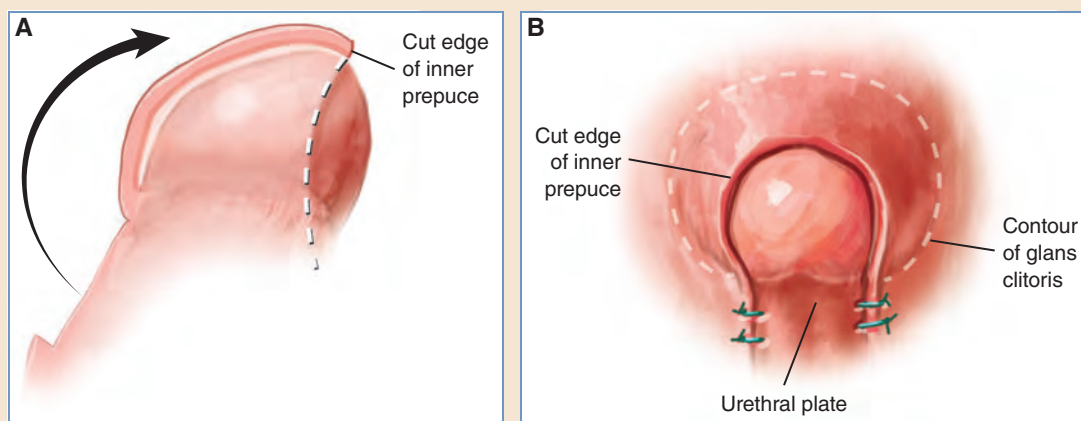


Fig. 62-10 Creation of the inner layer of the clitoral hood. The collar of the inner prepuce is flipped over to cover the glans clitoris. **A**, A sagittal view shows the shape of the hood. **B**, The hood is not cinched around the glans but instead will be sutured to the distalmost aspect of the urethral plate after the glans is recessed. The dotted line shows the contour of the glans clitoris beneath the inner layer of the hood. The glans has not been reduced but is well concealed.

pubis (Fig. 62-11). A large glans may be inadequately supported on the base of the clitoral body. It may tip more unnaturally toward the urethra and appear more conspicuous with retraction of the labia, particularly if the mucosal strip (former urethral plate) between the urethra and vagina is left too short. In this case, supporting sutures may be effective in the dorsal midline of the dermal side of the preputial hood, but they must not injure, impinge on, or kink the neurovascular tissue.

The remaining prepuce and clitoral skin are split in the midline, thus creating Byars flaps (Fig. 62-12). The skin is split approximately 1.5 cm from the base and adjusted as needed for the unique anatomy and glans size. The intact skin becomes the outer layer of the clitoral hood. The dermis is judiciously plicated to create a subtle finlike contour for the clitoral hood. The surgeon's eye determines the appropriate extent of contouring (Fig. 62-13). The outer hood is sutured to the inner hood with interrupted, fine absorbable sutures.

Labioplasty

Byars flaps, which are created from the sensate clitoral skin, become the labia minora. If no additional excess urogenital sinus tissue is available to line the vestibule, the medial edge of the Byars flap is sutured to the mucosal strip/urethral plate with interrupted absorbable sutures. Because the flaps are essentially rectangular and not representative of typical labia, judicious tapering and trimming distally will achieve the desired configuration. Similarly, a dog-ear of tissue is excised

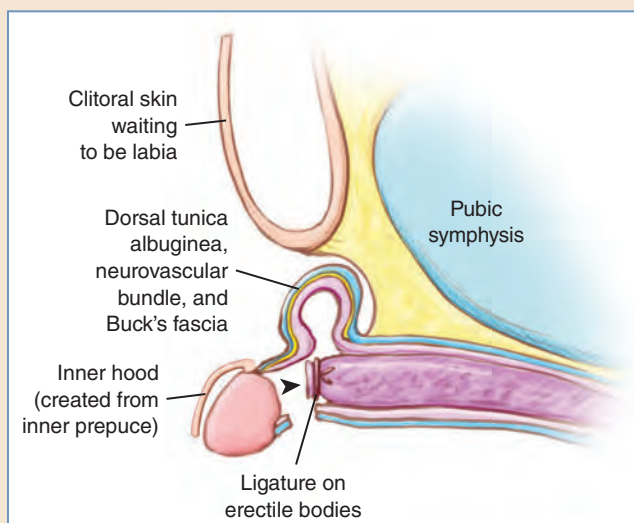


Fig. 62-11 A sagittal view as the glans is seated on the proximal erectile bodies. The erectile tissue has been ligated for hemostasis, but the tunica albuginea remains intact. The position of the dorsal tunica albuginea, neurovascular tissue, and Buck's fascia is shown.

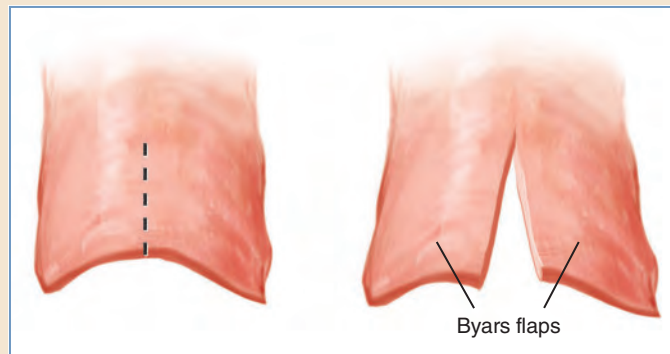


Fig. 62-12 Preparing the skin for the clitoral hood and labia minora. The remaining dorsal skin is split in the midline to create Byars flaps. The incision is stopped about 1.5 cm from the base to preserve intact skin for the outer layer of the clitoral hood.

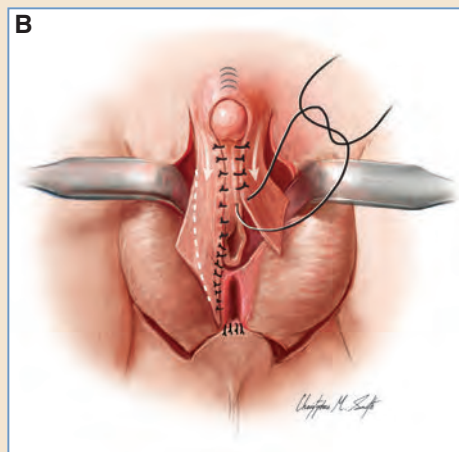
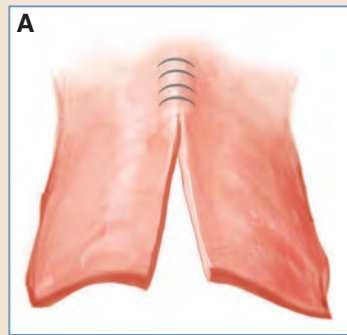


Fig. 62-13 Finishing the clitoral hood and creating labia minora. **A**, The outer layer of the hood is contoured by placing several absorbable sutures on the underside to plicate the dermis (the *caret marks* are shown to signify contour). **B**, The inner and outer layers of the hood are joined. Byars flaps flank the urethral plate and urethra or urogenital sinus to become the labia minora.

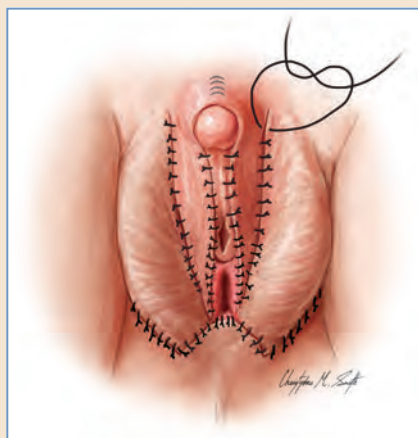


Fig. 62-14 Tailoring the labia minora. The shape of the Byars flaps is not representative of the labia minora, but as the tissue is tucked down, it develops a finlike contour that is not well represented in the two-dimensional view. The outer corner is trimmed for a tapered contour, and a dog-ear of tissue is excised anteriorly to create a contour between the labia minora and labia majora.

anteriorly between the inner and outer labia to create a natural-appearing cleft (Fig. 62-14). A two-layer closure with dermal anchoring sutures and cutaneous sutures is recommended at the tips of the labia minora to minimize the risk of dehiscence.

Cosmetic work on the labia majora will depend on whether urogenital mobilization and vaginoplasty with or without a posteriorly based perineal flap were undertaken during the reconstruction. The labia majora are mobilized by a U-shaped incision with the underlying fibrovascular and lipomatous tissue and moved posteriorly to flank the vaginal introitus. If an omega flap was advanced and incorporated into the posterior vaginal wall, the labia majora advance posteriorly, lateral to the omega flap. Debulking for a smooth contour may be incorporated for more virilized, rugated labia, although rugation cannot be fully addressed surgically. If urethroplasty and vaginoplasty were deferred, typically in patients with lesser labial fusion, reconfiguration of the labia majora would also be deferred in most patients.

OUTCOMES

From surgeons' perspectives, cosmetic outcomes have been considered very good for decades. We are, however, obligated to seek means to objectify cosmetic outcomes and to assess satisfaction and sexual function, because findings from the most contemporary clitoral reduction techniques are not yet available. Limited early information suggests a favorable outcome. However, measures to assess cosmetic outcome, intact neurovascular function, and adequacy of sexual function are not well defined.^{13,15} Intact perfusion and sensation have been physically confirmed in the first few years after nerve-sparing ventral clitoroplasty, but longer-term testing is needed.¹⁶ Fully intact sensation to touch and vibration may not be the only factor in perception of normal sexual function,¹⁷ but others find a linear relationship between sensation and function.¹⁸ Older reports describe results from historical techniques or very heterogeneous groups and may or may not be relevant.^{15,19} Nonanatomic (hormonal or psychological) factors may also affect sexual function.²⁰ We do not understand the impact of deferring surgical decision-making until adolescent–young adult age. More than one study suggests that patients prefer surgery at a young age, but the individuals queried had surgery in childhood.^{20,21}

After reconstructive surgery is performed in infancy or early childhood, it takes time to reach an age for confidence regarding intact sexual function. Relatively uncommon diagnoses yield relatively uncommon procedures and develop into meaningful uniform series only after many years. More work is needed in objectively assessing outcomes with and without surgical intervention.

KEY POINTS

- Patients with disorders of sex development, or intersex conditions, should receive short-term and long-term guidance of a multidisciplinary team of specialists. Long-term psychological support for the patient and parent is essential.
- Surgical management, including all elements of feminizing genitoplasty, should only be undertaken after extensive counseling and only by surgeons with a strong grasp of the anatomy and experience with complex genital reconstruction.
- Current understanding of genital anatomy indicates that nerve-sparing approaches to clitoral reduction are the best choice to preserve clitoral function. Additional physical testing and validated patient-reported outcomes are needed to fully understand clitoral function and sexual function outcomes after feminizing genitoplasty.
- Appropriation of tissue resources must be considered, whether a single-stage procedure with feminizing genitoplasty and management of the urogenital sinus/vaginoplasty is planned, or whether the latter portion will be deferred until maturation.

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63

Phalloplasty

Richard J. Redett • Sami H. Tuffaha



Without proper treatment, a male child with penile absence, deficiency, or deformity can undergo debilitating functional and psychosocial sequelae. Beyond the obvious impact on urinary and sexual functioning, a significant penile defect can damage a boy's developing sense of identity and self-esteem, with profound and long-lasting effects on global well-being and interpersonal relationships. Reconstructive surgery can have a strong, positive impact when integrated into a holistic team approach that addresses the urologic, sexual, and psychological pathologies associated with genital defects.

CONGENITAL ANOMALIES

Penile defects requiring surgical reconstruction can result from trauma, neoplasm, and vascular disease; however, congenital anomalies are the most common cause in pediatric patients.¹ The congenital anomalies most likely to warrant phalloplastic reconstruction include micropenis, aphallia, ambiguous genitalia, epispadias, and bladder and cloacal exstrophy (Table 63-1).

Each of these malformations is associated with a unique set of genital, urologic, and pelvic anatomic defects and physiologic abnormalities that must be treated. Knowledge of how each of these malformations affects normal anatomy and sexual and urologic function will facilitate surgical decision-making and patient counseling and will maximize the likelihood of positive outcomes.

Table 63-1 Common Penile Congenital Anomalies

Condition	Incidence	Definition	Etiologic Factors
Aphallia	1 in 10,000,000	Agenesis of the penis	Failure in embryologic development of the cloacal folds to generate genital tubercles
Micropenis	1 in 200	Proper proportions and development but <2.5 standard deviations below the appropriate size for age	Defect in the hypothalamic-pituitary-gonadal axis
Ambiguous genitalia	1 in 4200	Also known as <i>disorder of sex development</i> (DSD) A spectrum of disorders leading to questionable or ambiguous genital appearance at birth	Congenital adrenal hyperplasia Leydig cell failure Testosterone biosynthesis enzyme defects Androgen insensitivity syndrome Congenital anorchia Gonadal dysgenesis Klinefelter syndrome (47,XXY)
Hypospadias	1 in 250	Urethral malformation, early termination and opening on the ventral penis	Undervirilization, decreased testosterone or dihydrotestosterone, elevated antimüllerian hormone
Epispadias	1 in 120,000	Urethral malformation, early termination and opening on the dorsum of the penis	Failure of mesodermal migration with premature rupture of the cloacal membrane Failure of midline fusion of the genital tubercles
Bladder exstrophy	1 in 10,000 to 1 in 50,000	Abdominal wall defect, pelvic diastasis, penile defect, epispadias, bladder open and exposed	Failure of mesodermal migration with premature rupture of cloacal membrane Failure of midline abdominal wall fusion after separation of genitourinary and gastrointestinal tract
Cloacal exstrophy	1 in 200,000 to 1 in 400,000	Abdominal wall defect with extrusion of the hindgut and cecum and splitting of the bladder, penis, and scrotum	Failure of mesodermal migration with premature rupture of the cloacal membrane Failure of midline abdominal wall fusion before separation of the genitourinary and gastrointestinal tracts

A cursory understanding of genital embryogenesis is helpful in understanding the development of congenital genital anomalies. The human fetus is sexually undifferentiated at 8 weeks' gestation and has both wolffian (male) and müllerian (female) genital ducts. From 12 to 16 weeks' gestation, sexual differentiation occurs. The developing testes secrete antimüllerian hormone, resulting in regression of the müllerian ducts. Androgens released from the testes maintain the wolffian ducts, which give rise to the internal genitalia and allow differentiation of the external genitalia; this process involves enlargement of the genital tubercle into the glans, fusion of the urethral/genital folds to form the shaft, and midline fusion of the genital swellings to form the scrotum. Derangements in this process can lead to congenital anomalies of the genitalia.

Aphallia is a rare condition in which the penis fails to develop in utero, resulting in complete absence of the penis. Many of these patients have associated genitourinary anomalies that need to be addressed. Historically, these patients underwent gender reassignment surgery, including orchiectomy, at a young age. However, this practice has largely fallen out of favor, because evidence suggests that many of these patients will maintain a male identity.² Many are good candidates for phalloplasty.

The word *micropenis*, or *microphallus*, refers to a normally formed penis that is shortened more than 2.5 standard deviations below the age-adjusted mean. The cause of micropenis is often endocrinologic in nature, with insufficient androgen stimulation in utero. Patients with hypogo-

nadotropic hypogonadism have hypothalamic dysfunction resulting in insufficient gonadotropin-releasing hormone (GnRH) production, whereas those with hypergonadotropic hypogonadism have inadequate androgen release in response to a normal GnRH stimulus. The first-line treatment for micropenis is testosterone, either systemically or topically. However, in severe cases that do not respond to medical management, surgical lengthening and skin grafting of the penis or phalloplasty can be considered at a later age (Fig. 63-1).

The term *ambiguous genitalia* describes a broad set of congenital defects in which the external genitalia have both male and female characteristics. Micropenis is often grouped into this category. Ambiguous genitalia can develop in males and females; however, typically only genetic males with ambiguous genitalia are considered candidates for phalloplasty. Ambiguous genitalia in genetic males can have an array of features, including a small penis resembling a clitoris, a urethral opening above or below the penis, an undifferentiated scrotum resembling labia, and undescended testes. The specific surgical approach to ambiguous genitalia depends on which features are present (Fig. 63-2).

Hypospadias is a commonly occurring congenital penile defect in which the urethral meatus is proximal and ventral to its normal location in the distal glans. It is the result of partial or complete failure of the urethral folds to fuse, causing disruption of distal urethral formation.



Fig. 63-1 Two adolescent males with micropenis.

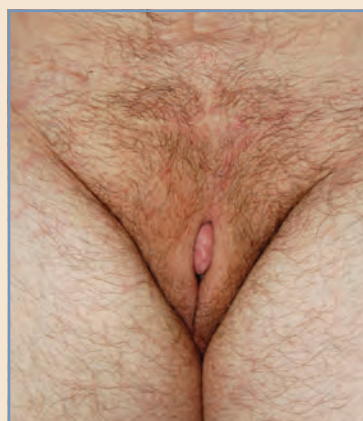


Fig. 63-2 This genetic male with ambiguous genitalia had early gender reassignment. He was raised as a female but later underwent gender reconversion to a male in adolescence.

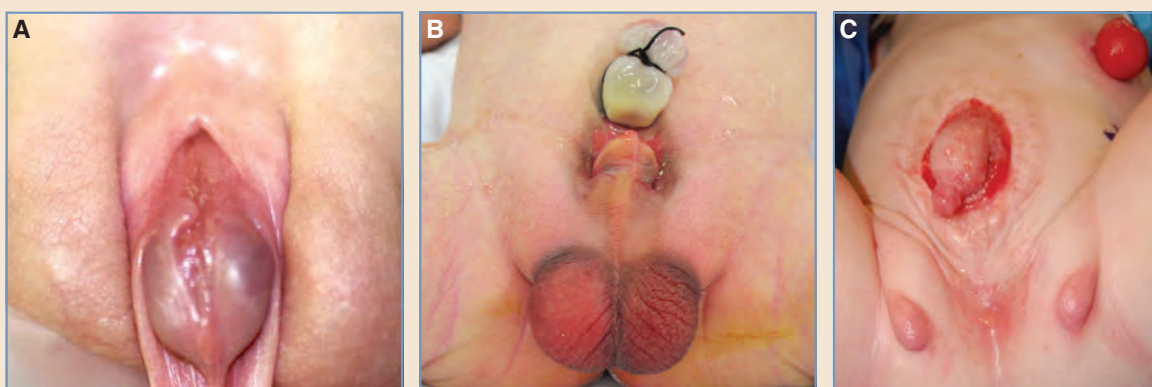


Fig. 63-3 Exstrophy-epispadias complex. **A**, This 6-month-old boy has epispadias. **B**, This 8-month-old boy has bladder exstrophy. **C**, This 12-month-old boy has cloacal exstrophy.

Epispadias, bladder exstrophy, and cloacal exstrophy are thought to belong to a spectrum of genital malformations called an *exstrophy-epispadias complex* (EEC).³ Bladder exstrophy presents with a lower abdominal wall defect and severe pubic symphysis diastasis, with exposure of an open bladder and urethra and dorsal urethral opening. Cloacal exstrophy is more severe, with a bilobed bladder separated by the cecum. Epispadias, the mildest form of EEC, presents with a closed bladder, a dorsally open urethral meatus, and mild pubic diastasis. These defects result from failure of mesodermal migration into the cloacal membrane that separates the ectoderm and endoderm of the anterior abdominal wall. This presumably results in premature rupture of the cloacal membrane. The timing and location of rupture determine whether a patient will develop epispadias, bladder exstrophy, or cloacal exstrophy.⁴

The genital defects associated with EEC can present a difficult reconstructive challenge. Management often involves staged reconstruction of the bladder, urethra, abdominal wall, and genitalia and requires close coordination and cooperation between the reconstructive surgeon and the urologist. The genital defects associated with EEC vary in severity. In isolated epispadias, the urethral meatus is located on the dorsal surface of the penis at the penopubic angle, and the glans is open dorsally. The phallus is typically shortened with dorsal chordee. Epispadias can sometimes be managed with standard lengthening procedures; however, phalloplasty can achieve favorable results in severe cases. In cases of exstrophy, the penis can be halved and separated from the midline. In such cases of severe deformity, phalloplasty is indicated^{5,6} (Fig. 63-3).

GENDER DYSPHORIA

Pediatric phalloplasty is typically performed to reconstruct anatomic penile defects only; however, reconstructive surgeons may see patients seeking gender reassignment. *Gender dysphoria* describes a marked difference between a person's expressed/experienced gender and the gender perceived by others. According to the Diagnostic and Statistical Manual of Mental Disorders (DSM-5),⁷ the condition must be present and stable for at least 6 months to be diagnosed. Gender dysphoria can manifest in many ways. For example, a patient can have strong feelings and reactions typical of the other gender and strong desires to be treated as the other gender and/or to change his or her own anatomic characteristics to those of the other gender. For patients suspected of having gender dysphoria, referral to appropriate specialists for multimodal therapy is critical.

A recent report by the American Psychiatric Association Task Force on Treatment of Gender Identity Disorder⁸ provided evidence-based treatment recommendations for patients with gender dysphoria. For children younger than 12 years of age diagnosed with gender dysphoria, psychoeducation and counseling to optimize psychological adjustment and well-being are the mainstays of treatment. Without treatment, most of these children will eventually become comfortable with their natal gender, and some will identify as transgender or transsexual in adulthood.⁹

In adolescents with persistent cross-gender identity and behavior, gender dysphoria often worsens with the onset of puberty. In addition to psychotherapy, education, and counseling, these patients may be appropriate candidates for hormonal therapy with gonadotropin-releasing hormone to suspend puberty. Before hormonal therapy is considered, coexisting psychological conditions that may contribute to dysphoria and confound an accurate diagnosis need to be identified and treated. In the United States, gender reassignment surgery is rarely, if ever, performed for patients younger than 18 years of age.

NONOPERATIVE AND OPERATIVE TREATMENT OPTIONS

Congenital penile deficiency resulting from fetal testosterone deficiency may respond to testosterone treatment delivered systemically and/or locally. A response to testosterone treatment can be seen in infancy, later in childhood, and in adolescence. Testosterone is the first-line treatment for patients with congenital hypogonadic micropenis. If medical treatment is unsuccessful, penile lengthening or phalloplasty can be considered.

Currently, congenital micropenis resulting from fetal testosterone deficiency is the only congenital penile defect that has a medical treatment. All others are treated with surgical intervention. Milder genital defects are sometimes amenable to primary repair, local tissue rearrangement, and/or lengthening procedures. Some defects are amenable to local flap reconstruction, but the surrounding tissue has been scarred by multiple previous operations. In these cases, tissue expansion has shown great utility.¹⁰ Tissue expansion is also useful in conjunction with lengthening procedures (Fig. 63-4). Tissue expansion can also be used to create a neourethra with capsular flags in the setting of hypospadias repair⁹ (Fig. 63-5). However, when a

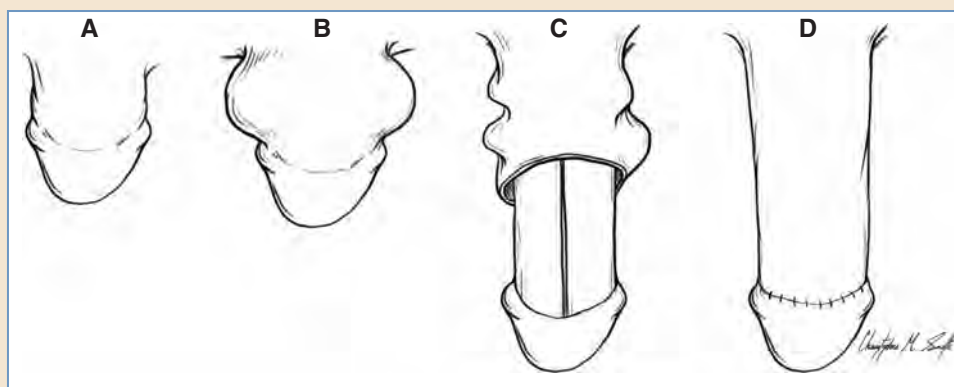
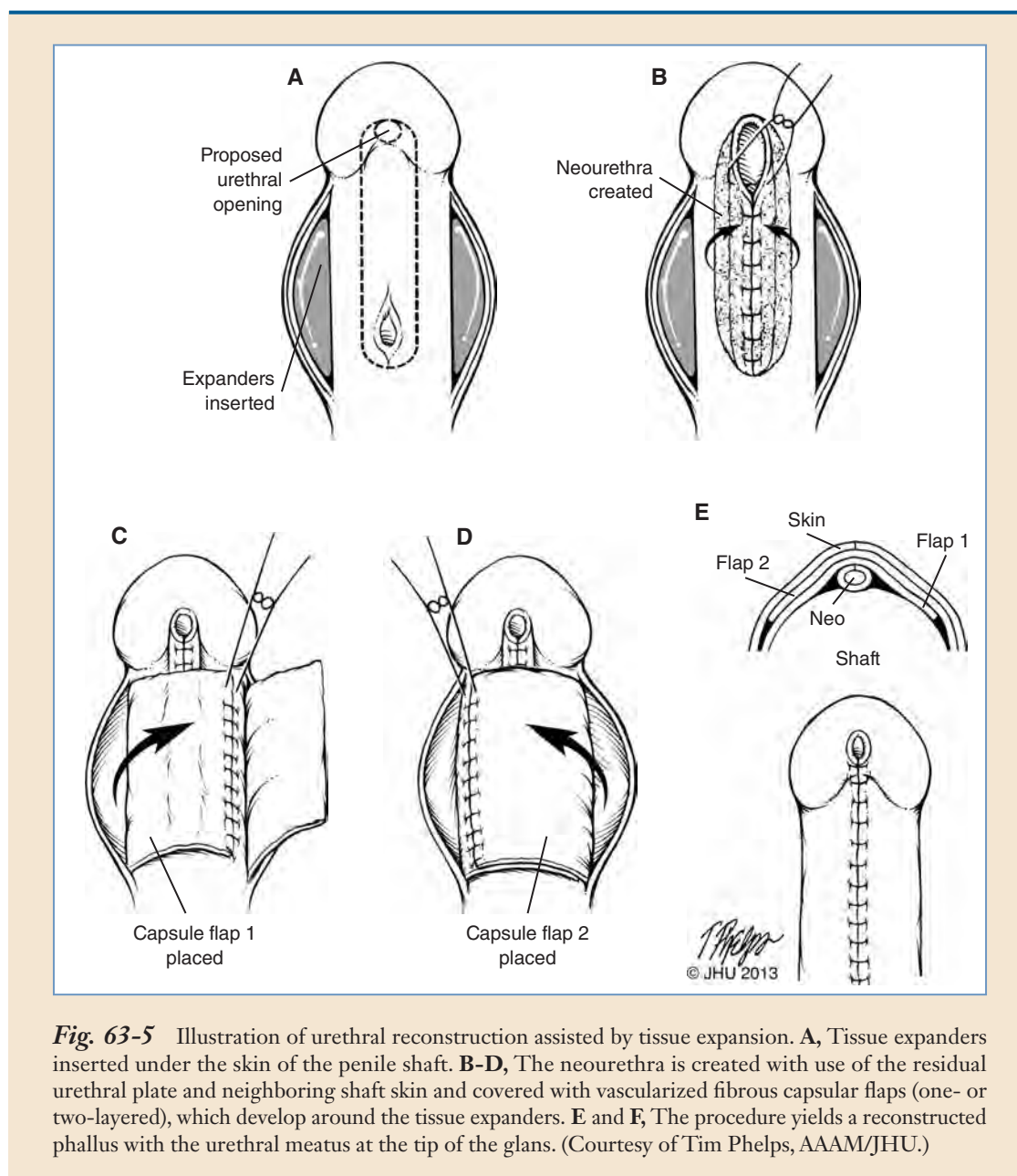


Fig. 63-4 A, Micropenis. B, Tissue expanders are inserted under the shaft skin bilaterally. C and D, When the tissue expander is removed, the underlying structures are lengthened, and the shaft is resurfaced with redundant skin.



satisfactory result cannot be achieved, phalloplasty with pedicled locoregional flaps or free tissue transfer should be considered.

In the past, aphallia and severe congenital penile defects were treated with gender reassignment, with bilateral orchiectomy, penectomy (when a penis was present), labioplasty, clitoroplasty, and vaginoplasty. This practice is no longer popular, because patients typically maintain a male identity after surgery, and effective techniques for phalloplastic reconstruction have emerged¹¹ (see Fig. 63-2).

Options for Phalloplasty

The goals of phalloplastic reconstruction include the formation of an aesthetically acceptable neophallus; a competent urethra that allows voiding while standing, in patients without continent urinary stomas; enough stiffness for sexual penetration; erogenous and protective sensation; and minimal donor-site morbidity. Over the past century, the surgical approach to total phalloplasty has evolved significantly. Numerous approaches have emerged, each providing some, but not all, of the ideal characteristics of a neophallus. The earliest techniques used pedicled flaps from the abdomen and groin. More recently, with the advent of microsurgical technique, a number of options involving free tissue transfer have gained popularity. Because they typically provide better function, sensation, and aesthetic outcomes, free flaps have largely replaced pedicled options for phalloplasty.

Pedicled Abdominal Flap

In 1936 Bogoras¹² described the first total penile reconstruction, which he called “penis plastica totalis.” He used a random-pattern, pedicled flap from the abdomen and reported sexual function. Taylor et al¹³ and Davies and Matti¹⁴ in 1988 described a single-stage approach to a pedicled rectus abdominis myocutaneous flap supplied by the deep inferior epigastric vessels. The flap was tubularized, and an implant was inserted, facilitating sexual function. Although the flap is a reasonable option when microsurgery is not possible, it is insensate, and the aesthetics are often suboptimal.

Pedicled Groin Flap

Smith et al¹⁵ first described the pedicled groin flap for phalloplasty in 1972. It is an axial flap based on the superficial circumflex iliac vessels and performed in a single-stage reconstruction, with or without an implantable device to provide stiffness. In 1990 Sun et al¹⁶ reported on a modification of the groin flap that incorporated a portion of the iliac crest to allow penetration. Although a groin flap is an option for phalloplasty, it is limited by poor sensation and aesthetics, similar to a pedicled abdominal flap. Furthermore, groin flaps often result in a small neophallus, because the donor tissue is limited. For these reasons, the groin flap for phalloplasty has largely been replaced by free flap options.

Radial Forearm Free Flap

Ever since Song¹⁷ described the radial forearm free (RFF) flap in 1982, its popularity has grown, and many consider it to be the benchmark for phalloplasty.¹⁸ RFF flaps are well suited for creating a neophallus, in part because it is relatively thin, supple, and hairless, thus mimicking native penile shaft skin. With additional modifications to define the penile subunits, the flap provides much-improved aesthetic outcomes (Fig. 63-6). Furthermore, because it receives robust sensory innervation from both the medial and lateral antebrachial cutaneous nerves, an RFF flap, when neurotized, can provide more than adequate erogenous and protective sensation, and patients often report the ability to orgasm.⁶ To achieve rigidity, an implant is often inserted in a second stage. However, the placement of an implant introduces a significant risk of extrusion.^{6,18} An alternative approach to provide rigidity without an implant involves harvesting a portion of the radius within the flap; however, this approach is limited by the nuisance of permanent rigidity and a high rate of bone resorption. RFF flaps are prone to high rates of urinary fistula and stricture and a conspicuous donor site that carries the stigma of gender reassignment in transsexual patients.^{19,20}

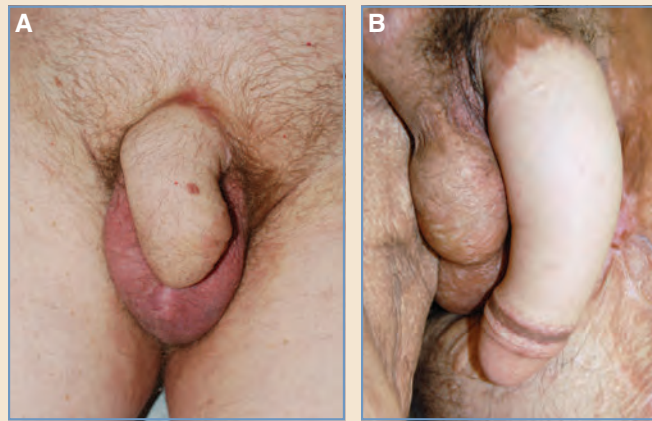


Fig. 63-6 A, An RFF flap without glanuloplasty and coronaplasty modifications. B, An RFF flap with glanuloplasty and coronaplasty modifications, resulting in improved aesthetics and a more realistic appearance of the neophallus.

Anterolateral Thigh Flap

Felici and Felici²¹ first introduced the anterolateral thigh (ALT) flap for phalloplasty in 2006. The flap is raised as a fasciocutaneous perforator flap based on the descending branch of the lateral circumflex femoral vessels and can be used as a free pedicled flap for phalloplasty. This flap has several theoretical advantages over an RFF flap, including a less conspicuous donor site, skin that is potentially less pigmented, and greater soft tissue bulk.²² However, the skin of an ALT flap is often much thicker and less supple than that of an RFF flap; therefore it may be less aesthetically pleasing. Similar to RFF flaps, ALT flaps can be neurotized for sensation using the lateral femoral cutaneous nerve of the thigh; however, it appears to provide significantly less erogenous sensation than that of RFF flaps.²³

Latissimus Dorsi Myocutaneous Free Flap

The latissimus dorsi myocutaneous free flap was first used for phalloplasty in 1996.²⁴ Based on the thoracodorsal vessels, this flap is a reliable option for phalloplasty that provides considerable soft tissue bulk. More recently, modifications to the flap for neurotization have been described. Perovic et al²⁵ reported that approximating the thoracodorsal nerve of the flap to the ilioinguinal nerve result in return of some protective sensation. However, the mechanism by which this occurs is unclear, given that a sensory nerve regenerates into a motor pathway. Even with neurotization, erogenous sensation and orgasm have not been achieved with the use of latissimus dorsi myocutaneous free flaps. Ranno et al²⁶ approximated the thoracodorsal nerve of the flap to the nerve of the gracilis and reported that patients were able to voluntarily contract the latissimus muscle within the flap to achieve penetration without an implant.

Fibular Osteocutaneous Free Flap

To overcome the lack of intrinsic rigidity of the RFF flap and the resulting need for an implant, Sadove et al²⁷ in 1993 reported using a fibular osteocutaneous free flap for phalloplasty. The flap is perfused with the peroneal vessels, and the lateral sural cutaneous nerve can be used to provide sensory neurotization, although the sensibility of the flap is poor compared with that of an RFF

flap. The primary advantage of fibular osteocutaneous free flaps is inclusion of a long portion of the fibula to facilitate penetration without an implant. Despite the value of intrinsic rigidity provided by this flap, a permanently erect neophallus can be difficult to conceal and uncomfortable. Furthermore, the bony component within the flap is highly susceptible to resorption and warping.

PREOPERATIVE ASSESSMENT AND PLANNING

The preoperative assessment of a patient with a congenital penile defect begins with a careful physical examination and discussion with the referring urologist to determine the extent of the defect. During the initial evaluation, surgeons should consider whether a reasonable result can be achieved with lengthening procedures and local flaps or skin grafts or whether neophallus reconstruction is required to provide an acceptable outcome. The components of the native penis that can be incorporated into a neophallus are identified. If a functional glans is present, it can be incorporated into the ventral base of the neophallus to augment erogenous sensation. When the corpora are present and salvageable, they can also be incorporated to provide some soft tissue bulk and stiffness within the flap and to help seat an erectile implant. However, in cases of aphallia or severely deformed genitalia, incorporating native components may not be an option.

The length and patency of the urethra and the status of the more proximal urinary system are important preoperative considerations. The goal is to allow urination through the neophallus when possible. However, many children with bladder or cloacal exstrophy undergo bladder neck transection and creation of an umbilical continent urinary stoma. The native urethra is brought out through the ventral base of the neophallus to allow ejaculation, but patients will continue to catheterize the umbilical urinary stoma after reconstruction.

Surgeons need to determine whether associated deficits and comorbidities will preclude or minimize the usefulness of phalloplastic reconstruction. The reconstruction options are carefully explained to the patient's caregivers. Older children and teenagers are included in these conversations and play a central role in decision-making. Setting realistic expectations for patients and families is critically important.

After a decision is made to proceed with phalloplasty, the various donor-site options are thoroughly discussed with the patient and family. The potential donor sites are examined to assess appropriateness for use. For example, patients with excessive subcutaneous adiposity may not be ideal candidates for trunk- and thigh-based flaps. Previous surgical scars are another reason to avoid a particular donor site. Before proceeding with an RFF flap, an Allen test and Doppler ultrasound of each extremity are always performed to ensure adequate circulation to the hand through the ulnar artery. If these tests are equivocal, angiography may be performed.

When deciding on the optimal timing for phalloplasty, surgeons should consider the potential for neophallic growth and whether the penile construct will contain somatic and/or androgen-sensitive tissue components. A patient's native penis will grow exponentially during puberty in response to androgen surges. However, a neophallus constructed from somatic tissue will undergo much more modest growth. Therefore a patient's donor site must be large enough to construct a neophallus of nearly adult proportions. There are psychological advantages to performing phalloplasty in younger, prepubescent patients. For many surgeons, total penile reconstruction is best performed in patients 10 to 14 years of age. Surgery at this time minimizes the psychosocial distress that results from penile deformity and allows patients to adapt to the neophallus at a younger age. The growth patterns and psychological maturity of each patient needs to be considered when selecting the optimal timing for reconstruction. We have found that the likelihood of success is maximized when reconstruction is delayed until patients can fully cooperate with surgery and postoperative care. Most patients are able to do this after entering puberty.

SURGICAL APPROACH TO THE RADIAL FOREARM FREE FLAP FOR PHALLOPLASTY

Given the superior aesthetics and sensibility provided by RFF flaps, many consider it to be the benchmark for phalloplasty, and it is the senior author's (R.J.R.) preferred option. The surgical approach for an RFF flap will be described in detail, but many of the technical aspects and general considerations are applicable to the other approaches for phalloplasty (Figs. 63-7 through 63-10).

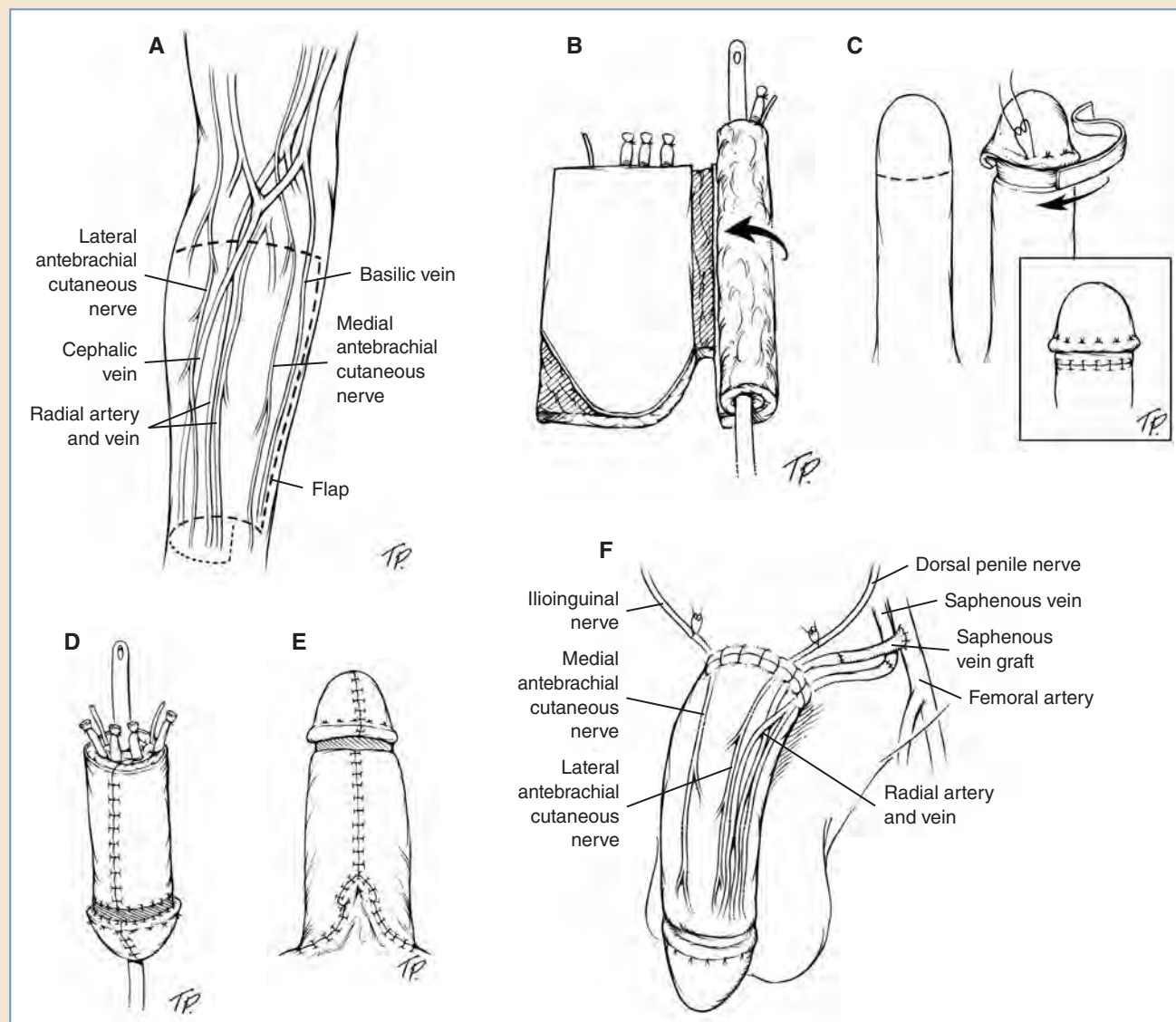


Fig. 63-7 The key steps of RFF flap phalloplasty. **A**, The flap is outlined, with important neurovascular structures to be included. **B**, The flap is rolled from ulnar to radial. An ulnar skin flap is rolled over itself to create a neourethra. The central, deepithelialized skin is buried in the flap. The flap is then rolled under itself to externalize the radial skin. **C**, Glanuloplasty and coronoplasty. The skin flap is folded inward and sutured to itself to create a glans. A full-thickness skin graft is applied to the donor site of the glans flap to create the appearance of a circumcision scar at the corona. **D**, The complete, prelaminated flap before transfer. **E**, The native glans is inset into the ventral base of the neophallus. **F**, The transferred flap, showing the vascular anastomoses and nerve coaptations. (Courtesy of Tim Phelps, AAAM/JHU.)

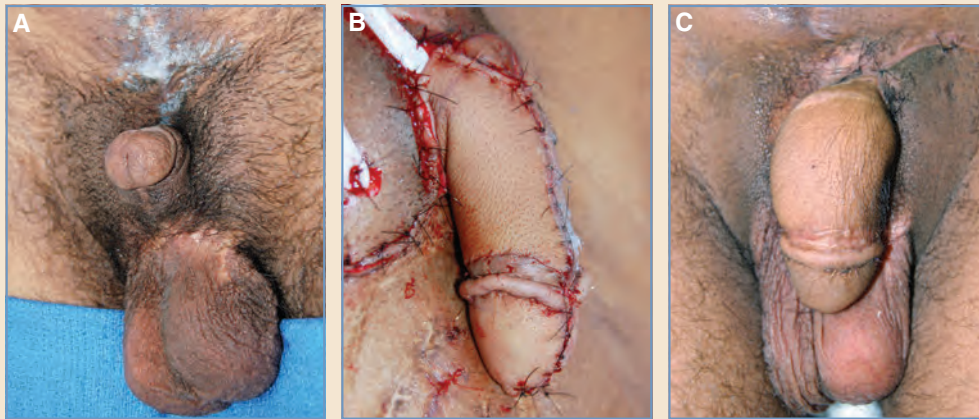


Fig. 63-8 A, This male patient with bladder exstrophy and micropenis is shown preoperatively. B, The intraoperative appearance of a neophallus created with an RFF flap phalloplasty. C, The postoperative result.

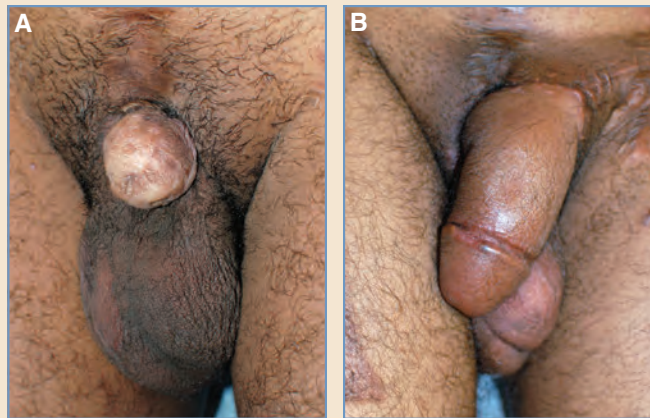


Fig. 63-9 A, This male patient had bladder exstrophy. B, He is shown after reconstruction with an RFF flap phalloplasty.

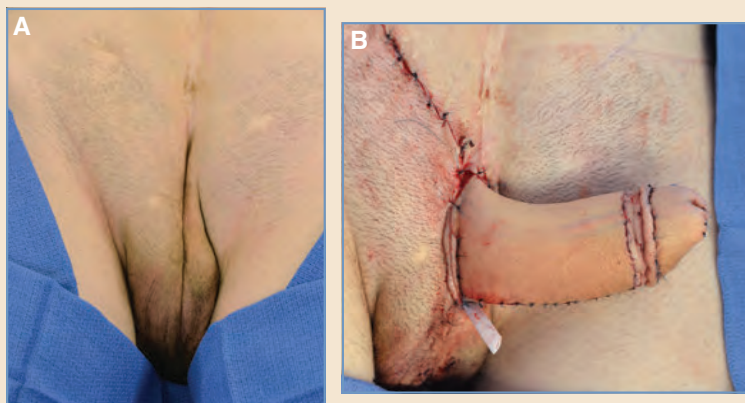


Fig. 63-10 A, This male patient had severe cloacal exstrophy and ambiguous genitalia. B, He is shown after reconstruction with an RFF flap phalloplasty.

Preparation and Harvest

The patient is placed in a supine position with the upper extremity from which the flap will be harvested abducted and supinated. A sterile upper extremity tourniquet is used during flap harvest for hemostasis. The flap is marked with dimensions of 14 by 14 cm. This typically includes the entire width and up to three fourths of the length of the volar forearm, beginning distally at the proximal wrist crease. Before the flap is elevated, the neoglans is defined by raising a transverse split-thickness skin flap across the distal forearm and rolling it under itself, where it is secured with resorbable sutures. The donor site of the glans flap is later covered with a full-thickness skin graft to better define the corona and to give the penis the appearance of having a circumcision scar. A coronal glans flap can also be created once the neophallus is tubed, but we think it is easier to do so before the flap is harvested. For patients who require a neourethra, a central longitudinal strip of skin is deepithelialized, separating the ulnar-sided skin panel (which will line the neourethra) and the radial-sided skin panel (which will provide the external skin of the neophallus).

The RFF flap is then raised in a standard fashion from distal to proximal. The radial artery and venae comitantes are identified between the flexor carpi radialis and the brachioradialis at the wrist and ligated. These vessels are elevated with the flap from the underlying tendons, while preserving the paratenon. We include the palmaris longus tendon, which is used to anchor the neophallus to the pubic bone. The medial and lateral antebrachial cutaneous nerves and the cephalic vein are identified and harvested with the flap. The flap is tubularized on the forearm before the pedicle is transected. From ulnar to radial, the flap is first rolled over itself to form the neourethra with the ulnar-sided skin panel and then under itself to externalize the radial-sided skin panel. The deepithelialized central panel is buried within the flap.

Recipient Site Preparation and Flap Inset

If two surgical teams are available, the recipient site and flap can be prepared simultaneously. A pediatric urologist is typically present to assist with recipient site preparation.

If the native penis is to be incorporated into the neophallus, the shaft skin is removed, and internal structures, including the urethra and the penile nerves, are isolated. When a competent urethra in continuity with the proximal urinary tract is present, the corpus spongiosum is severed from the glans so that it can be anastomosed to the neourethra within the flap. When the bladder neck has previously been transected, and the patient voids from an umbilical stoma, the urethra is left in continuity with the glans, and the external meatus is externalized through the ventral base of the neophallus to allow ejaculation. In either case, the glans is incorporated into the neophallus. It is first deepithelialized and then buried into the base of the neophallus, either during the phalloplasty or at a later stage to preserve a lifeboat in case of flap failure.

The dorsal penile nerves are isolated for erogenous sensation, and either the ilioinguinal or iliohypogastric nerve is isolated for protective sensation. These are approximated to the medial antebrachial cutaneous and lateral antebrachial cutaneous nerves. A number of recipient site vessels can be anastomosed to the radial artery to perfuse the flap, including the superficial circumflex iliac, the inferior epigastric, and the external pudendal and femoral arteries. The accompanying veins and the great saphenous vein can be used to provide venous outflow through the venae comitantes of the radial artery and the cephalic vein. We perform as many venous anastomoses as possible to reduce postoperative swelling and venous congestion.

The neourethra within the flap is anastomosed to the native urethra over a 10 or 12 Fr urinary catheter at the phalloplasty procedure. This is the senior author's preferred method for neourethra construction. Alternative approaches include prelaminating the flap with a full-thickness skin graft and inserting a tubed skin or buccal mucosa graft at a later stage.

Penile Prosthetic Implant

A penile prosthetic implant is inserted at a later stage and is typically delayed until late adolescence or early adulthood to minimize implant-associated risks, including soft tissue erosion, infection, and extrusion. Before an implant is inserted, protective sensation should be present and the patient should have a high level of maturity. We wait a minimum of 1 year after phalloplasty before inserting an implant to ensure the return of protective sensation before the procedure.

OUTCOMES AND LIMITATIONS

Many surgical options are available for phalloplasty, but none provides all of the qualities that a neophallus should have, including entirely acceptable aesthetics, a competent urethra with low risk of fistula and stricture, tactile and erogenous sensibility, enough stiffness and durability to allow sexual penetration, and minimal donor-site morbidity. An RFF flap is the most popular option for phalloplasty, because it provides excellent aesthetics and consistent erogenous and protective sensation. However, as with all methods of phalloplasty, the flap has significant limitations that require serious consideration. All of our patients who underwent RFF flap phalloplasty reported the ability to achieve orgasm and satisfaction with the neophallus appearance. Roughly half of our patients have had significant complications involving urinary strictures and fistulas and/or implant-associated complications requiring implant explantation; these findings are consistent with other large series.^{6,18}

FUTURE TREATMENT MODALITIES

Promising potential alternatives on the horizon may address the limitations of autologous phalloplastic reconstruction. For select patients in whom traditional reconstruction was unsuccessful and those without usable donor sites, a penile transplantation may offer a solution. For these difficult cases, the benefit provided by penile transplantation may outweigh the risks associated with immunosuppression. Careful screening, informed consent, and psychological evaluation will be of utmost importance in selecting appropriate patients, and the pool of eligible recipients will be limited and exclude the pediatric population. However, if and when tolerance protocols are developed that obviate the need for lifelong immunosuppression, penile transplantation may supplant traditional reconstruction, given the potential for markedly improved functional and aesthetic outcomes without donor-site morbidity.

Before penile transplantation can be successfully implemented, a number of technical considerations need to be addressed. We recently performed cadaveric perfusion studies to determine the optimal approach to vascularizing penile allografts. Because of the complexity of penile vascular anatomy, a number of vascular pedicles will have to be included to optimally perfuse the many crucial components of a penile allograft. Although the dominant blood supply to the penis is provided by the dorsal arteries, anastomosing the cavernosal arteries will probably augment corporal inflow necessary for engorgement and erection. For proximal levels of transplantation, blood supply from the external pudendal system may be important to prevent skin necrosis^{28,29} (Fig. 63-11 and Table 63-2).

Perhaps farther on the horizon, regenerative medicine may provide an ideal solution for penile reconstruction. Recent successes have been made in implanting tissue-engineered subunits of the genitourinary tract in animals and humans.³⁰ However, the complexity of penile anatomy presents many hurdles to overcome before a complete, functional penis can be grown in a laboratory.

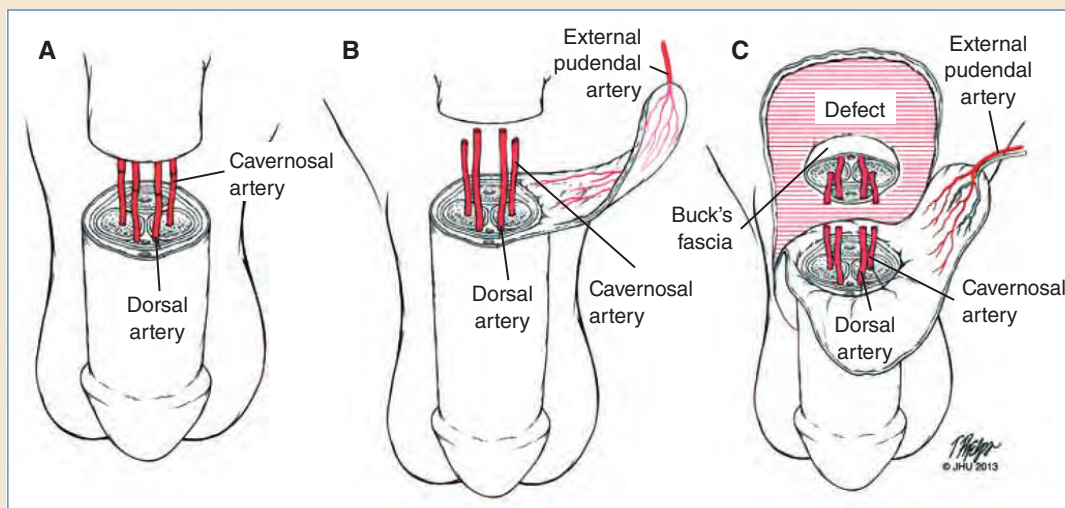
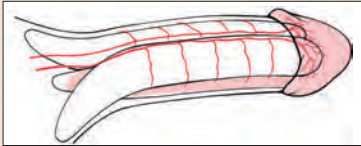
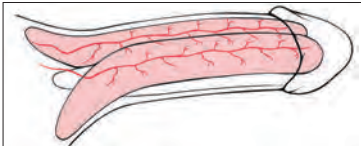
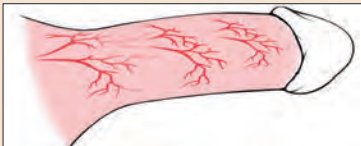


Fig. 63-11 A recommended strategy for vascularizing penile allografts for reconstructive transplantation. **A**, Midshaft or distal shaft transplant: dorsal and cavernosal arteries only. **B**, Proximal shaft: dorsal, cavernosal, and external pudendal arteries with a skin bridge. **C**, The proximal shaft with the surrounding defect: dorsal, cavernosal, and external pudendal arteries with additional skin to resurface the defect. (Courtesy of Tim Phelps, AAAM/JHU.)

Table 63-2 Perfusion Territories

Artery	Principle Perfusion Territory	
Dorsal	Glans and corpus spongiosum	
Cavernosal	Corpus cavernosum	
External pudendal	Shaft skin	

Courtesy of Tim Phelps, AAAM/JHU.

KEY POINTS

- Congenital anomalies are the most common cause of penile defects requiring reconstruction in pediatric patients.
- Genetic males with aphallia or ambiguous genitalia should be considered for phalloplasty; early gender reassignment surgery for these patients has fallen out of favor, because many patients maintain their male identity.
- Congenital penile anomalies resulting from fetal testosterone deficiency may respond to testosterone therapy.
- *Gender dysphoria* is a term describing a marked difference between a person's expressed/experienced gender and the gender others would assign to him or her.
- Gender reassignment surgery is typically not performed before patients are 18 years of age.
- Some penile defects can be corrected with tissue expansion, local tissue rearrangement, and lengthening procedures.
- The goals of phalloplasty include the formation of an aesthetically acceptable neophallus; a competent urethra for voiding while standing, in patients without a continent urinary stoma; enough stiffness to allow sexual penetration; erogenous and protective sensation; and minimal donor-site morbidity.
- We consider the radial forearm free flap to be the benchmark for phalloplasty, because it provides superior aesthetics and sensation.
- Tissue engineering and vascularized composite allotransplantation may offer improved results in the future.

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Anastomosing the cavernosal arteries may augment corporal inflow, which is necessary for erection. Although the dorsal arteries are critical for distal penile skin perfusion, the external pudendal artery should be used in proximal transplantation to ensure adequate shaft skin perfusion. Each of these arteries has a distinct and seemingly important perfusion territory that should be considered in a penile transplantation.
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Pressure Sores and Neural Tube Disorders in Children

Michael S. Gart • Gregory A. Dumanian • Russell R. Reid



In the mid-1800s, Brown-Séquard¹ proposed pressure as the cause of bedsores in paraplegic patients. Since then, pressure sores have posed a significant challenge to the medical community. Plastic surgeons often lead the multidisciplinary team management of these wounds. Although both adult and pediatric patients develop these wounds, the underlying causes and comorbidities, treatments, and treatment success are significantly different. Adult patients have high recurrence rates of up to 80%,²⁻⁶ whereas pediatric patients have recurrence rates as low as 5%.⁷ This chapter focuses on the epidemiology, etiologic factors, conservative and operative management, and outcomes of pediatric pressure sores. Relevant data from our institution are presented to support the potential long-term success of operative repair in pediatric patients.

EPIDEMIOLOGY

Despite increasing awareness of the economic and medical consequences of pressure sores,⁸ their development is a persistent, common, and formidable challenge for health care institutions. In the United States, the incidence of this disease entity approaches 40% in some hospitals, 25% in long-term nursing facilities, and 20% in home care settings.⁹ The average cost to heal a single pressure sore has been estimated to range from \$500 to \$78,000, with the total burden on the health care system exceeding \$1.3 billion annually.¹⁰ Medicare will only reimburse for the care of stage III and IV pressure ulcers (see Table 64-1) that are documented on patient admission.¹¹ With average annual hospital costs of \$400,000 to 700,000¹² related to pressure ulcer management, adequate prevention strategies have significant financial implications.

The incidence and location of pediatric pressure ulcers vary with the study population. In a study of pediatric intensive care units, Curley et al¹³ found that 199 pressure sores developed in 86 of 322 patients (27%) on bed rest for at least 24 hours. Most of these sores (70%) were stage I; of the 60 stage II and III ulcers, 19 (32%) involved the head. In contrast, Baldwin¹⁴ conducted a national survey in which the incidence of pressure ulcers was 0.29%, and the prevalence was 0.47%. At our institution (Shriners Hospitals for Children—Chicago), from 1995 to 2004, 641 patients presented with spinal cord injuries. In 126 patients (20%) a pressure ulcer was documented at some point during this 9-year span; in 48 patients (7%) a pressure sore was the primary reason for admission. Based on this data set, the incidence of patients with spinal cord injury admitted to our institution over the study period was 0.8%.¹⁵

Some of the underlying causes leading to the formation of pressure sores are unique to the pediatric population. Although traumatic causes of paraplegia are common denominators in the development of both adult and pediatric pressure sores, children have congenital causes such as myelodysplasia, spina bifida, and/or myelomeningocele. These entities will be discussed in brief detail in the chapter. In contrast, adults more often have cerebrovascular accidents and chronic, debilitating neurologic illnesses.

Moreover, the anatomic distribution of pressure sores consistently differs between adults and children. In adults the most common sites are the ischium (28%), trochanter (19%), sacrum (17%), calcaneus (9%), and other sites (27%).¹⁶ In children, however, the relatively large size and weight of the head leads to a higher incidence of occipital pressure sores, and the use of lower extremity casting and orthotics increases the incidence of sores in the lower extremity. In children with an uncorrected kyphoscoliotic spine, pressure sores tend to develop anywhere along a bony prominence, which may lie in the cervical, thoracic, and lumbar region. Children with significant kyphoses also have problems with sitting because of altered loading of the pelvis and ischial tuberosities.

ETIOLOGIC FACTORS

The final common pathway in the development of pressure sores is the compression of soft tissues resulting in ischemia. If this pressure is not relieved, the eventual outcome is necrosis and ulceration. Although pressure is the single most important factor in the occurrence of these wounds, other local and systemic factors contribute; these include infection, malnutrition, obesity, insensate areas, immobility, paralysis, incontinence, and impaired cognition.¹⁷

Microinjection studies have demonstrated that systemic blood pressure is transmitted to the capillary level, with a mean transcapillary gradient of 20 to 40 mm Hg and a mean capillary pressure of 32 mm Hg.¹⁸ If compression between a bony prominence and a firm surface exceeds the capillary perfusion pressure, tissue ischemia results. In most people, ischemia is relieved by postural changes or by removal of external compression, and ischemia is reversible; however, in critically ill patients and those unable to change position or sense pressure, ischemia may progress to tissue necrosis. The exact duration of ischemia that results in ulceration is determined by the interplay of multiple factors, including tissue susceptibility, shearing forces, the amount of pressure, and the length of time it is applied.

The relationship between time and pressure for developing pressure sores was originally studied in animals¹⁹ (Fig. 64-1). Kosiak et al^{19,20} found that 70 mm Hg applied over a 2-hour period was sufficient to cause microscopic evidence of ischemia in dogs. Daniel et al^{21,22} confirmed these findings and in studies showed that 500 mm Hg of compression applied for 2 hours or 100 mm Hg applied for 10 hours was sufficient to cause muscle necrosis. Skin necrosis did not occur until 600 mm Hg was applied for 11 hours, suggesting that skin is more tolerant of ischemia than muscle. This is clinically significant for two reasons: First, sitting individuals have been shown to generate pressures as high as 500 mm Hg in the buttock region, and second, the highest pressures are generated deep, adjacent to pressure points such as bone, with less pressure at the

skin level.¹⁸ Skin is more resistant to ischemia, and the early visible changes in the skin may not reflect the actual injury in the muscle and deeper tissues. Therefore the cutaneous changes in a pressure sore have been described as the tip of an iceberg. Furthermore, Dinsdale^{23,24} demonstrated that friction can decrease the amount of pressure required to produce ulceration.

In a classic study, Lindan et al²⁵ placed patients on a compressible bed of springs and nails to measure the distribution of pressure in supine and sitting positions. Pressures of 40 to 60 mm Hg were recorded in the sacrum, heels, and buttocks while patients were supine, whereas pressures of up to 100 mm Hg were recorded in the ischium of patients in the sitting position²⁵ (Fig. 64-2).

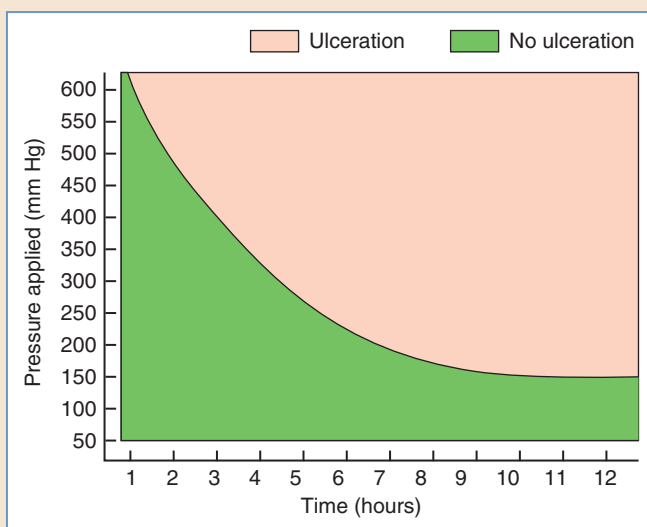


Fig. 64-1 The inverse relationship between time (hours) and pressure (mm Hg) in the pathogenesis of pressure sore formation.

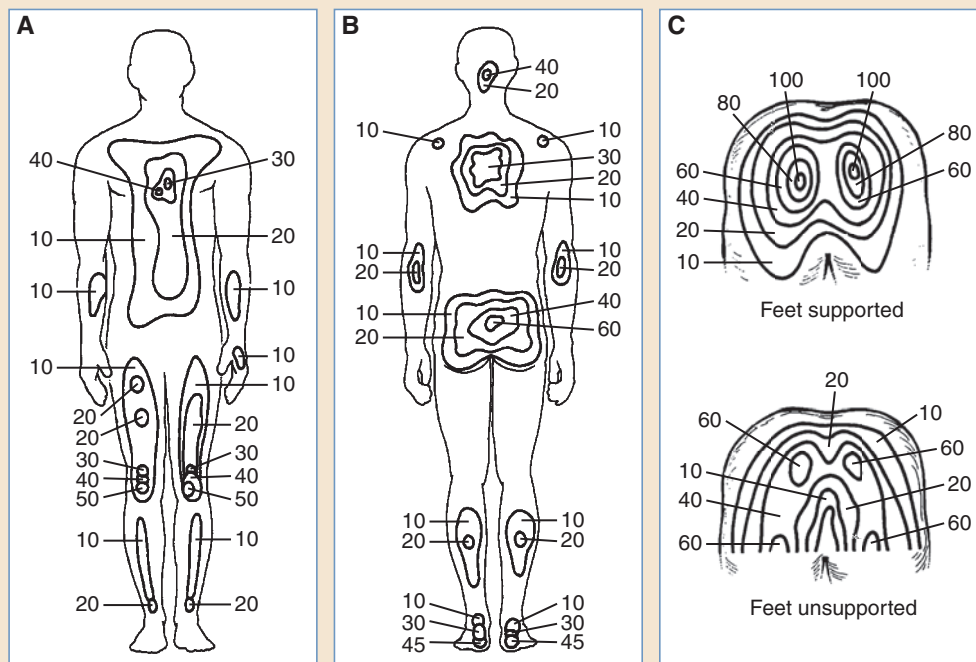


Fig. 64-2 Distribution of pressure (mm Hg) in a healthy adult. A, Prone. B, Supine. C, Sitting.

NEURAL TUBE DEFECTS

Neural tube defects (NTDs) are the second most common congenital defects, after congenital heart defects, and affect approximately 0.5 to 1 per 1000 established pregnancies worldwide.²⁶ The clinical severity of this class of defects varies greatly from uniformly fatal in utero (anencephaly and craniorachischisis) to asymptomatic (spina bifida occulta). The most common group of central nervous system anomalies that is compatible with life is spina bifida, a failure of vertebral arch formation that results in a bifid vertebral column. The most frequently observed form of spina bifida is myelomeningocele, in which the spinal cord extrudes into a meninges-covered sac filled with CSF. Despite preventive strategies, including folic acid supplementation, the incidence of myelomeningocele in the United States has stabilized at 3.4 per 10,000 births.²⁷ Myelomeningocele is generally compatible with life; however, neurologic impairment is seen below the level of the lesion and can result in numerous neurologic conditions, including paraplegia. Neural tube defects and their resulting paralysis/insensibility are a common reason pediatrics patients present with pressure sores. Moreover, these patients may present in the immediate postpartum period to plastic surgeons for closure of the myelomeningocele defect. In this situation, evidence suggests that local and regional flaps can be used to provide layered closure with good outcomes.²⁸ Fetal surgery has addressed these defects prenatally,²⁷ although currently, this treatment option is rare because of limited access and the dangers of fetal surgery.

PREOPERATIVE ASSESSMENT

History and Physical Examination

The diagnosis of pressure sores is relatively straightforward and based largely on the underlying disease process and a thorough examination of the wound. Various staging systems have been developed to facilitate serial examinations and communication among medical personnel. The National Pressure Ulcer Advisory Panel Consensus Development Conference²⁹ developed the most widely used system in 1989 and redefined it in 2007 (Table 64-1). However, such systems are subject to interrater variability and may not accurately describe wounds that appear superficial, but extend into the deep tissues.

All wounds should be examined for fluctuance, which may suggest a deep fluid collection that requires operative drainage. All necrotic tissue must be thoroughly debrided before initiating conservative or operative therapy. For insensate patients, this may be initiated at the bedside; however, definitive debridement is best performed in an operating room, where lighting and equipment for bony debridement and hemostasis are optimal. Osteomyelitis may be clinically suggested by a soft character to palpable bone at the base of a pressure ulcer; however, histopathology analysis provides the definitive diagnosis. The presence of osteomyelitis needs to be determined, because these wounds are more prone to breakdown and recurrence if not adequately debrided and treated with an appropriate course of antibiotics, usually prescribed by an infectious disease consultant.

Laboratory Tests and Imaging Studies

Several laboratory parameters, including erythrocyte sedimentation rate (ESR) and C-reactive protein (CRP) have been suggested to screen for the presence of osteomyelitis; however, these tests have poor specificity. The use of MRI to diagnose pressure sore-related osteomyelitis has been reported, with a sensitivity of 98% and a specificity of 89%.³⁰ However, in a 3-year follow-up, Huang et al³⁰ found that, despite 86% (49 of 57) positive MRI readings, only 48% of patients (20 of 42) required surgical debridement. The authors concluded that this resulted from the tendency for MRI to “overcall” the existence of osteomyelitis. Other imaging modalities, including

Table 64-1 Pressure Sore Stages

Stage	Title	Description
I	Nonblanchable erythema	Intact skin with nonblanchable erythema over a localized area, usually over a bony prominence, that lasts for more than 1 hour after pressure relief May be painful May indicate people at risk
II	Partial-thickness	Partial-thickness loss of dermis presenting as a shallow, open, red-pink ulcer without slough May also appear as an open or ruptured blister Should not be used to describe skin tears, tape burns, incontinence-associated dermatitis, maceration, or excoriation
III	Full-thickness loss, skin loss	Full-thickness tissue loss Subcutaneous possibly visible, but not bone, tendon, or muscle Depth varies by anatomic location
IV	Full-thickness tissue loss	All soft tissue destroyed, with exposed, directly palpable bone, tendon, or muscle, making osteomyelitis or osteitis likely
Additional categories	Unstageable/ unclassified Suspected deep tissue injury	Full-thickness skin or tissue loss of unknown depth: completely obscured by slough and/or eschar in wound bed Depth unknown: purple or maroon discoloration of intact skin or blood-filled blister caused by damage of underlying tissue from pressure and/or shear

Source: National Pressure Ulcer Advisory Panel (NPUAP), NPUAP Pressure Ulcer Stages/Categories. Available at <http://www.npuap.org/resources/educational-and-clinical-resources/npuap-pressure-ulcer-stagescategories/>.

CT and Tc⁹⁹ bone scanning have demonstrated limited utility in diagnosing osteomyelitis, with a sensitivity and specificity of 11% and 64% and 57% and 90%, respectively.³¹ CT and/or MR imaging may be useful in confirming deep fluid collections and/or sinus tracts that may not be apparent during surgical debridement. In a landmark paper, Lewis et al³¹ established histopathology as the benchmark for diagnosing osteomyelitis. They performed a Jamshidi core needle bone biopsy of 52 patients and reported a sensitivity and specificity of 73% and 96%, respectively.

NONOPERATIVE TREATMENT

Many stage I and II pressure sores can be treated with preventive or conservative measures aimed at halting progression of the wound and improving the local environment to maximize healing.

Preventive Management

The Braden Scale for Predicting Pressure Sore Risk is an instrument for pressure sore risk assessment that was developed for adults but is often used in children. It is a summated rating scale with scores ranging from 1 to 4 on five scales, and 1 to 3 on a friction and shear scale³² (Table 64-2). The best treatment for pressure sores is prevention, and this scale helps to alert the medical team to the potential risk of pressure sore development in immobilized patients. Because the single most important etiologic factor in the development of these wounds is pressure, relief of pressure is tantamount to prevention. Frequently, despite this information and application of meticulous prevention strategies, pressure sores develop. Therefore an aggressive multidisciplinary

Table 64-2 Braden Scale Components

	Sensory Perception	Moisture	Activity	Mobility	Nutrition	Friction and Shear
1 =	Completely limited (unresponsive)	Constantly moist	Bedfast	Completely immobile	Very poor	Problem
2 =	Very limited (responds to painful stimuli)	Very moist	Chairfast	Very limited	Probably inadequate	Potential problem
3 =	Slightly limited (responds to verbal stimuli)	Occasionally moist	Walks occasionally	Slightly limited	Adequate	No apparent problem
4 =	No impairment	Rarely moist	Walks frequently	No limitation	Excellent	

This is a summated rating scale with score possibilities ranging from 1 to 4 on the first five scales, and 1 to 3 on the friction and shear scale. The lower the score, the higher the ulcer risk potential.

approach is required that includes teaching patients and caregivers how to adequately relieve pressure and promptly recognize early skin changes.

Wound Care

The tenets of wound care are not specific to patient age, which has led to the extrapolation of adult treatment strategies to pediatric patients. For outpatient care, either air, foam, gel, or a water overlay/mattress supporting 19.5 kg/m³ is recommended for low-risk patients and should be immediately replaced with an air-exchange mattress at the first signs of hyperemia on a pressure point.³³ Similarly, all moderate- to high-risk patients should have a dynamic air-exchange mattress in the outpatient setting and be placed immediately on such a mattress on admission to the hospital.³³

Pressure sores are chronic wounds that incite a catabolic state. Nutritional supplementation is initiated to meet the increased protein and energy requirements to heal a pressure sore. Protein supplementation of 1.5 g/kg/day has been shown to not only improve the overall morbidity in these patients but also to enhance prevention of sore formation.^{34,35} Nutritional consultation and calorie counts should be implemented in all moderate- to high-risk patients.

Regardless of the pressure sore stage, we follow the surgical principle of moist wound healing.^{36,37} We often find that before a plastic surgical consultation, the initial approach to wound management was to “dry out” the wound, which tends to compromise tissue barriers and potentiate infection.

With recent advances in wound care products, myriad dressing choices are available to maintain a moist healing environment. These can be broadly classified as hydrocolloids and alginates, enzymatic agents, antimicrobials, and negative pressure wound therapy (NPWT). Characteristics of hydrocolloids (that make them favorable for localized care of pediatric pressure sores) are their ability to absorb exudative fluid, semipermeability, adhesiveness, and relatively infrequent need for dressing changes (every 3 to 5 days). Reducing the frequency for potentially painful dressing changes improves compliance and favors healing. These absorptive dressings should not be used for wounds with active infection, wounds with exposed tendon and/or bone, and

patients with diabetic foot ulcers. Although these dressings are costly, compared with some alternatives, the reduced need for dressing changes makes them cost-effective, compared with traditional gauze/saline-moistened dressings. Other disadvantages of these dressings include potential maceration of wound margins, limited wound visibility, and the foul odor that they produce.³⁸ Calcium derivatives of alginic acid, alginates, have absorptive qualities and the ability to solubilize and form hydrophilic gels in the presence of exudative wounds.³⁹

Recently, enzymatic debriding agents have been increasing in popularity. Ointments containing collagenase are effective in wounds with minimal necrotic debris (less than 20% of the total wound surface) *when used in conjunction with sharp debridement*. These enzymes successfully digest proteins from nonviable tissue, resulting in smaller peptides that are eliminated from the wound. We have found that long-term use of this ointment can result in the development of hypertrophic granulation tissue, which can subvert epithelialization of the wound.

When a wound has been successfully debrided and freed from necrotic and infected tissue, topical antimicrobial agents may be applied for local wound care. Silver sulfadiazine (Silvadene) and mafenide acetate (Sulfamylon) are topical antibiotics whose predominant application is in burn care. Similar to burns, pressure wounds are relatively avascular, and circulating systemic antibiotics often do not reach therapeutic levels at the target tissues when given parenterally. Topical antimicrobial agents provide high local concentrations while limiting systemic absorption and potential side effects.

Silvadene is the most widely used topical antimicrobial in the United States, largely for its ease of use, broad antimicrobial coverage, and relatively limited toxicity. It is relatively painless on application, but contains sulfa moieties; therefore it cannot be used in sulfa-allergic patients. Moreover, Silvadene can cause neutropenia with prolonged use, requiring close monitoring in patients with immunocompromised states. It can cause kernicterus in infants younger than 2 months of age and should not be used in these patients.

Sulfamylon has excellent penetration into avascular tissues, including cartilage and eschar. It has activity against *Pseudomonas aeruginosa*; however, adverse effects, including painful application, metabolic acidosis, and high osmolality that desiccates the wound tissue, limit the use of this agent to specific situations.

Bacitracin and mupirocin are topical antimicrobial ointments that provide a moist healing environment, usually with a low adverse-effect profile. So-called “triple” antibiotic ointments can occasionally cause a local rash from skin sensitivity. In such cases, we typically recommend vitamin A&D ointment to maintain moisture in the wound.

NPWT, referred to as *wound VAC therapy*, creates a negative pressure environment in which interstitial edema is decreased, thereby improving local circulation and angiogenesis and enhancing the formation of granulation tissue.^{40,41} Several reports have demonstrated a role for NPWT in the healing of pressure sores. Philbeck et al⁴² showed that closure of deep wounds with NPWT (0.23 cm²/day) is faster than with saline-moistened gauze dressings (0.090 cm²/day)⁴³ for treating trochanteric and trunk sores in conjunction with a low-air-loss bedding system. NPWT also reduced dressing-change frequency. NPWT devices should not be placed on contaminated or infected wounds. For patients with systemic signs of infection, the device is removed to allow a full examination of the underlying wound.

SURGICAL TREATMENT

Surgical Preparation of the Wound Bed

Unlike stage I and II sores, more advanced (stage III and IV) pressure sores usually require surgical intervention: debridement, reconstruction, or both. Determining when a wound is unresponsive to conservative therapy and may need flap reconstruction is a major decision. There

is no clear answer to this question. Several factors must be considered before making such a treatment decision, including the patient's wishes, comorbidities, nutritional status, and level of compliance with treatment; the location and characteristics of the wound; and the philosophy of the treating surgeon. At our institution, we consider a wound amenable to flap reconstruction when nutritional deficiencies and other medical comorbidities have been addressed and it has failed to respond further to conservative measures. Patients need to be motivated and compliant with the postoperative pressure-relief protocol to protect the newly treated pressure site and to prevent the development of a new sore.

Conway and Griffith⁴⁴ aptly described the basic tenets of the surgical management of pressure sores as follows:

- Excision of the ulcer, surrounding scar, heterotopic calcification, and underlying bursa
- Partial or complete osteotomy to remove the bony prominence
- Closure of the wound, which includes filling dead space with fascia or muscle flaps and resurfacing it with healthy tissue that can provide durable coverage over the wound

Adequate debridement that removes all devitalized tissue is a mainstay in treating pressure sores, whether they are to be reconstructed or allowed to heal by secondary intention. In select patients, including insensate paraplegics and quadriplegics, adequate debridement may be performed at the bedside; however, the bleeding risk and lack of available instruments limits the adequacy of such a procedure, and we reserve this option for minor wounds or poor anesthetic candidates. When possible, we prefer to debride in the operating room, where anesthesia, positioning, instrumentation, lighting, and assistance are optimized. This is particularly true for pediatric patients who tend to tolerate bedside procedures more poorly than adults. The appropriate anesthetic for a given debridement depends on several factors, including the patient's level of sensation, the positioning requirements, the anticipated length of surgery, and the potential need for blood products. We find that pressurized water irrigation (pulsed lavage) and hydrosurgical debridement (VersaJet, Smith & Nephew) are useful adjuncts that have been shown to reduce bacterial contamination.⁴⁵ Furthermore, Dorafshar et al⁴⁶ described the technique of methylene blue-guided debridement, which can provide visual confirmation of adequate debridement.

Intraoperatively, tissue cultures should be sent to determine the microbial pathogens that inhabit a wound and their antibiotic resistance patterns. We routinely send samples before and after debridement to determine adequacy of debridement and the presence of residual infection. Some centers use intraoperative quantitative cultures to ensure acceptable bacterial loads before reconstructing a wound or completing debridement. We do not routinely perform such cultures, because clinical assessment of a contracting, granulated wound tends to suffice. If osteomyelitis is suspected clinically, pathologic specimens of bone can be analyzed for the presence of osteomyelitis. At our institution, debridement and reconstruction are staged to facilitate assessment of the bacterial colonization and to obtain an infectious disease consultation for managing osteomyelitis, when present, before the definitive reconstruction. In the interim between the debridement and the reconstruction, the conservative measures mentioned previously are initiated to maintain a clean wound until the flap is closed.

Removal of Bony Prominences

The need to debride or excise bony prominences in an established pressure sore is derived from the pathophysiology that leads to wound formation. A broader bony prominence will better distribute the forces applied to the soft tissues, thereby helping to prevent a new sore from developing. Moreover, excision of bone will help to improve the wound excision and bacterial clearance and will decrease tissue requirements for wound closure.³¹ However, overly aggressive resection of bone can lead to complications, which include skeletal instability, bleeding, and redistribution

of pressure to adjacent areas with subsequent development of new sores. For example, a unilateral ischiectomy significantly increases pressure on the contralateral ischium and can increase the occurrence of contralateral pressure sores. Similarly, a bilateral ischiectomy increases perineal pressure and leads to difficult perineal wounds and, in extreme cases, urethrocuteaneous fistulas.⁴⁷ Therefore bilateral total ischiectomies should not be performed. In cases of recurrent unilateral ischial sores, the spine and tilt of the pelvis should be analyzed as the inciting cause of the sitting imbalance and may require corrective spine surgery.

Removal of the proximal femur for the treatment of trochanteric ulcers is sometimes necessary, especially if the joint is involved.⁴⁸ After a Girdlestone arthroplasty, the dead space of the acetabulum should be eliminated with fascia or muscle. A vastus lateralis flap is most commonly used for this purpose. For cases of chronic hip subluxation with an associated pressure sore, the acetabulum is typically already filled with chronic scar tissue, and a flap to fill the dead space is no longer necessary. Wound closure is aided if movement at the distal femur, which occurs as a result of removal of the proximal femur, is minimal. This movement can be controlled by various techniques, including external fixation devices, external traction devices, and abduction pillows supplemented with antispasmodic drugs.^{48,49}

SURGICAL TECHNIQUE

Certain principles of flap closure of all types of pressure sores are applicable for pediatric and adult patients. Dead spaces should be filled. Skin closures should be performed with acceptable tension. Flaps should maintain pulsatile blood flow after they are moved into position and should be reelevated for recurrences. Finally, flaps should be planned to keep the newly placed incisions off of the expected area of maximal pressure.

Sacral Pressure Sores

The surgical management of sacral pressure sores spans the reconstructive ladder and ranges from undermining and primary closure to free tissue transfer (Box 64-1). Tissue loss resulting from the ulcer may result in primary closure under tension and a significant recurrence rate. Higher on the reconstructive ladder, skin grafting has been attempted, but has shown poor outcomes because of fragility of the soft tissue coverage over pressure-bearing areas. Therefore the most common reconstructive procedures for sacral pressure sores are myocutaneous and fasciocutaneous flaps after debridement and coccygectomy.

Box 64-1 Reconstructive Options for Sacral Pressure Sores

- Primary closure
- Skin graft
- Turnover gluteus myoplasty
- Gluteus maximus myocutaneous flap*
- Gluteus maximus fasciocutaneous flap
- Sensory island flaps
- Intercostal island flaps
- Perforator flaps (superior gluteal)
- Free tissue transfer

*The most commonly used option at our institution.

The workhorse flaps for sacral pressure sore reconstructions are based on the gluteal vascular system and may be designed as muscular, myocutaneous, or fasciocutaneous flaps. Furthermore, they can be based superiorly or inferiorly; part or all of the muscle can be included in the design; and they may be advanced, rotated, or turned over. We commonly place nonambulatory patients in the prone position and make markings for a unilateral V-Y advancement myocutaneous flap (Fig. 64-3). After the skin is incised, dissection through the gluteus maximus muscle begins at the superior and lateral border near the trochanter. With a finger in the interspace between the gluteus maximus and medius, the lateral portion of the muscle toward the insertion is divided, followed by division of the inferior portion of the muscle under the lower skin incision. The muscle is elevated in a medial-to-lateral direction off of the sacrotuberous ligament, the main palpable landmark of the procedure. With the muscle completely mobilized laterally and inferiorly, the two vascular pedicles are identified, facilitating the final division of the muscle off of its medial attachments to the sacrum. With the flap completely dissected and the vascular pedicles identified, the wound can be thoroughly debrided without inadvertently injuring the pedicle or pedicles. This flap can easily provide enough coverage of the sacrum without tension and places the final incision line off the midline pressure point. In patients with very large wounds, this may be performed bilaterally; the only downside is that midline placement of the incision can increase susceptibility to breakdown in the long term. Closed-suction drainage and postoperative pressure-relief beds are an integral part of all pressure sore reconstructions.

Other available flap reconstruction choices include perforator flaps, fasciocutaneous flaps, and innervated flaps. Recently, perforator flaps based on the superior gluteal artery have received

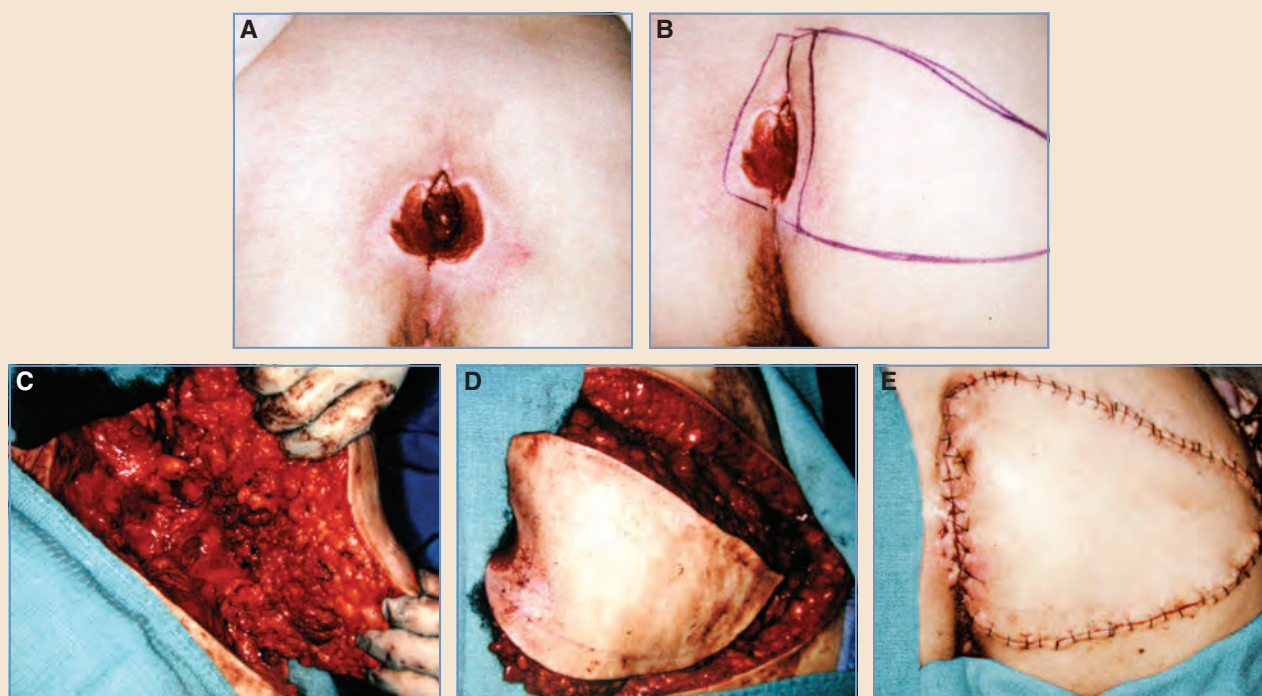


Fig. 64-3 Gluteus maximus V-Y myocutaneous flap for reconstruction of a sacral pressure sore. **A**, Sacral pressure sore. **B**, A unilateral V-Y advancement myocutaneous flap based on the gluteus maximus is marked. **C**, The flap is raised. **D**, The flap is inset without tension. **E**, The wound is closed in layers over drains.

attention for pressure sore reconstruction, especially for ambulatory patients.^{47,48} The main advantage of these flaps is preservation of the gluteus maximus muscle.

Neural Tube Defects

Duffy et al⁵⁰ have used perforator flaps for coverage of myelomeningocele defects, with no short-term recurrences. Some investigators have performed fasciocutaneous flaps because of the often highly atrophic nature of the gluteal muscle in this patient population. Proponents of this approach argue that regardless of the muscle atrophy, the increased soft tissue bulk of the muscle may protect against recurrence of sores. Evidence in animal models suggests that myocutaneous flaps have improved outcomes in contaminated wounds.⁵¹⁻⁵³ Innervated flaps have been developed to restore protective sensation. These techniques include tissue expansion of sensate skin more cranial on the back, intercostal flaps to bring sensation to the sacrum, and the use of innervated free flaps.⁵⁴⁻⁵⁷ Despite the theoretical advantage of this approach, the clinical utility of innervated flaps is limited.

Ischial Pressure Sores

As with sacral pressure sores, several techniques are available for treating ischial pressure sores (Box 64-2), most of which are myocutaneous flaps. These include flaps based on the gluteus maximus, hamstring, tensor fascia lata, and gracilis muscles. Flaps based on the gluteus may be superiorly or inferiorly based and can carry skin from the gluteal or thigh areas. The inferior gluteus myocutaneous flap is based on the inferior gluteal artery and uses only the inferolateral portion of the muscle. In this procedure, anesthesia is induced, and the patient is placed in the prone position. After adequate debridement and a partial ischiectomy are performed, the gluteus muscle is mobilized and divided from its inferior and lateral attachments to allow medial movement and obliteration of the potential space over the ischium. The muscle is inset and the skin closed in layers over closed-suction drains.

The hamstring comprises the biceps femoris, semimembranosus, and semitendinosus muscles. Flaps based on the medial thigh skin have the advantage of bringing in padded tissue and the option of readvancement if needed for recurrences. The usual design is a V-Y myocutaneous advancement flap. The procedure is performed with the patient in the lateral decubitus or prone position. After the cutaneous incisions are made, the attachments of the short head of the biceps femoris are released, preserving the perforating segmental branches from the femoral artery.

Box 64-2 Reconstruction Options for Ischial Pressure Sores

- Primary closure
- Gluteus maximus myocutaneous flap*
- Hamstring myocutaneous flap*
- Tensor fascia lata myocutaneous flap
- Gracilis myocutaneous flap*
- Anterolateral thigh flap
- Rectus abdominis myocutaneous flap
- Free tissue transfer

*The most commonly used options at our institution.

Next, the long head of the biceps femoris tendon is divided from its insertion onto the fibular head. Similarly, the distal insertions of the semimembranosus and semitendinosus muscles are divided. A skin bridge is preserved medially. The entire hamstring unit is elevated off of the femur, attached only by segmental branches of the profunda femoris vessel. The flap is then advanced superiorly to close the defect in a tension-free manner. The wound is closed in layers over drains.

The tensor fascia lata can be used to close ischial ulcers, but the distal end of the flap, which is used to fill the wound, may have unreliable vascularity. In appropriate situations, however, this flap can be designed as a sensate flap to provide protective sensation with the lateral femoral cutaneous nerve.⁵⁸⁻⁶⁰ In a series of patients with pressure sores treated with an extended, sensory tensor fascia lata flap, no new or recurrent pressure sores were observed after 1 to 10 years of follow-up.⁵⁹

The gracilis muscle is supplied by the medial circumflex femoral artery. With the patient in a prone position, an incision is made in the posteromedial aspect of the leg and continued until the gracilis and adductor longus muscles are identified. Three aids to identifying the gracilis muscle are as follows⁶¹:

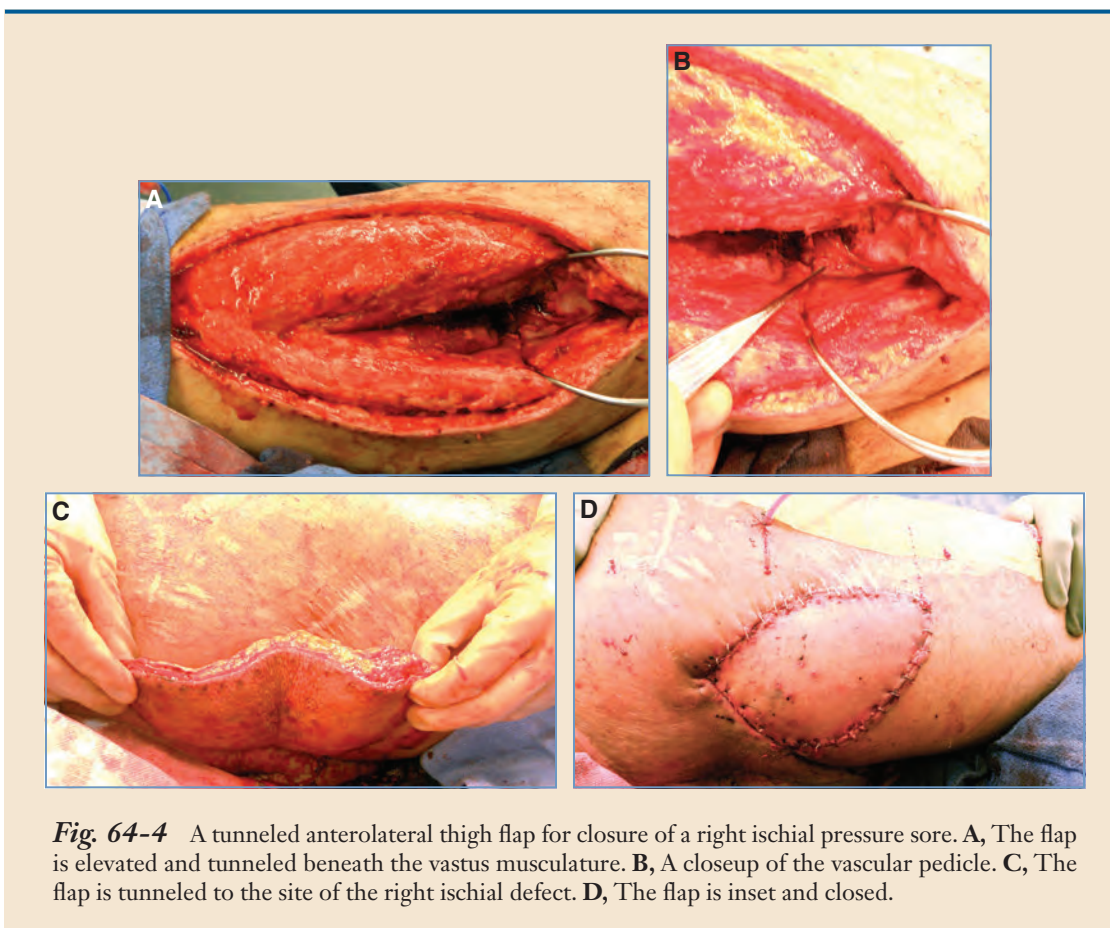
1. No superficial sensory or motor nerves are encountered during the dissection.
2. The dissecting finger can slide up and down the muscle on the deep and superficial surfaces without difficulty.
3. The muscle begins to taper and becomes triangular and tendinous two thirds of the distance down the thigh.

Once the muscle is identified, a skin paddle is designed and centered over the muscle. To prevent avulsion of the perforators, a suture is placed from the fascia of the skin island to the gracilis muscle. The dominant pedicle is exposed on the deep medial surface. After the gracilis muscle is detached from its tibial insertion, the flap is elevated from distal to proximal. If possible, the saphenous vein is kept in continuity proximally to improve venous outflow. Arterial inflow to the skin paddle is improved by including the adductor fascia with the flap. Flap movement is improved with division of the origin. After adequate debridement and a partial ischiectomy, the flap is inset. The gracilis flap has the advantage of being a true island flap, moving easily to cover the defect without tension and without a suture line over the central pressure point. The most significant disadvantage with this flap is occasional unreliability of the skin paddle.

An anterolateral thigh flap can be used for coverage of ischial sores by tunneling beneath the vastus muscle complex or subcutaneous tissues^{62,63} (Fig. 64-4).

Trochanteric Pressure Sores

Trochanteric pressure sores develop in patients who lie in the lateral decubitus position or sit in a significantly slouched posture in a wheelchair for prolonged periods of time. The risk is increased in patients with significant hip flexion contractures. As described previously, a Girdlestone arthroplasty with a vastus lateralis muscle flap should be considered if the hip joint is involved with the wound. However, a femoral head resection causes altered weightbearing on the proximal femur and ischium, and should not be performed if possible. Of the many reconstructive options for closure of trochanteric sores, the most commonly used are the tensor fascia lata or vastus lateralis myocutaneous flaps (Box 64-3). A tensor fascia lata flap can be transferred as muscle only, as muscle and skin, as an island flap, or as a free flap. It has the potential for sensation by the lateral femoral cutaneous nerve (L1-3) if the level of the spinal cord injury is below L3. Dissection begins at least 10 cm from the proximal knee to ensure good vascularity of the tip of the flap. Doppler signals are located on the skin paddle, and the skin incisions are made. The incisions are deepened through the fascia lata, and the pedicle is identified as it emerges from



Box 64-3 Reconstructive Options for Trochanteric Pressure Sores

- Tensor fascia lata myocutaneous flap*
- Vastus lateralis myocutaneous flap*
- Gluteus maximus or medius myocutaneous flap
- Free tissue transfer

*The most commonly used options at our institution.

under the rectus femoris to enter the tensor fascia lata muscle. The rest of the flap elevation off of the vastus lateralis is straightforward. The donor site is closed in V-Y fashion, primarily, or with a skin graft for large flaps.

The vastus lateralis is supplied by the descending branch of the lateral circumflex femoral artery and is the largest of the quadriceps muscle group, extending from the proximal femur to the patella. The flap can be raised as muscle only or as muscle and skin. The patient is placed in the lateral

decubitus position, and an incision is made on the anterolateral thigh from a point 10 cm below the anterior superior iliac spine to the lateral condyle of the femur. If a skin paddle is to be included, the incisions should be made at the borders of the island. The vastus lateralis is then identified and raised from distal to proximal. Care is taken to visualize and preserve the dominant pedicle that travels underneath the rectus femoris and not to elevate the vastus intermedialis (thereby exposing the femur). The flap is rotated into the wound and closed in layers over drains.

Scalp

Scalp pressure sores are usually stage I and II sores, as demonstrated by loss of hair follicles. These sores are seen more often in pediatric patients because of the larger relative size and weight of the head in this age group. Pressure sores of the scalp typically close with local wound care, consisting of pressure relief, cleaning with soap and water, removal of hair in the region close to the wound, and the application of topical antimicrobial agents. Surgical intervention is rarely required for these wounds, but when necessary, local rotation or advancement flaps are usually sufficient.

POSTOPERATIVE CARE

Flap closure of a pressure sore is one component of a comprehensive care plan that is essential for preventing postoperative complications, most notably dehiscence, partial or total flap necrosis, and recurrence. The factors that contributed to the original formation of the sore must be recognized and modified, and the patients and caregivers need to be informed about prevention. Optimizing nutrition and addressing medical comorbidities that may interfere with wound healing (diabetes and smoking) are critical. Furthermore, we employ a standard postoperative protocol at our institution that is aimed at pressure relief and a gradual return to activities of daily living.

The timing and degree of postoperative care regimens vary from one institution to another, but they all have in common pressure relief (by frequent transfers or special mattresses such as a Clinitron bed [Hill-Rom]), wound toilet to prevent maceration of the operative site, adequate drainage to remove serous fluid, and often antibiotics (based on the intraoperative cultures). At Shriners Hospitals for Children–Chicago, the following protocol is initiated after wound flap closure:

- The child is immediately transferred from the operating table to a Clinitron bed.
- Drains are placed on bulb suction and monitored for output, and these drains are kept in place for an average of 14 days.
- The medical staff meticulously cares for the skin, and the wounds are assessed for breakdown, scabs, drainage, and erythema.
- After 14 days in a Clinitron bed, a sitting protocol is initiated on an air mattress (Flexicair bed; Hill-Rom) (Table 64-3).
- If the surgical site is healing properly during the sitting protocol, the patient is allowed to sit with proper pressure-relief devices and pressure-relief methods (such as bending forward and elevating the buttocks, or turning while supine).
- Children are typically discharged 2 to 3 weeks after flap closure, with the sitting times explicitly listed for the patient and caregivers.
- Follow-up visits are scheduled within 2 weeks of discharge and monthly until the wounds are stable and healed.

Table 64-3 Sitting Protocol After Pressure Sore Flap Reconstruction

Week	Postoperative Day	Protocol
1-2	1-14	Bed rest
3	15-18	Sit 20 min QID
	19-21	Sit 40 min QID
4	22-24	Sit 60 min QID
	25-28	Sit 90 min QID
5	29-35	Sit 120 min
6 and beyond	36+	Sit maximum of 2 hr QID followed by 60 min of pressure relief until cleared by operating physician

QID, Four times a day.

TREATMENT OUTCOMES

Operative Complications

Acute complications after pressure sore debridement and reconstruction are similar to those of any surgical procedure and include pulmonary and cardiac complications, wound dehiscence, infection (wound and urinary tract), and hemorrhage. With sound surgical technique, many of these complications can be prevented; the main complication after pressure sore reconstruction is recurrence. Some studies have reported recurrence rates of up to 70%, although most of these studies have been performed in adult populations.⁶⁴ Because of the differences in underlying causes and medical comorbidities, the rate is probably much lower in the pediatric patients. A study from the Children's Hospital of Philadelphia reported a 5% recurrence rate in a series of 20 pressure sores.⁷ A 9-year retrospective review at Shriners Hospitals for Children–Chicago revealed a recurrence rate of 10%, with a follow-up ranging from 1 to 7 years.¹⁵ Regardless of the method of treatment, we think that key steps in reducing pressure sore recurrence are adequate preventive pressure-relief strategies; routine health care visits for evaluation and early detection of compromised soft tissue; and education of patients and caregivers.

Complications of Chronic Wounds

Chronic wounds are associated with several long-term sequelae. In pressure sores with underlying osteomyelitis, secondary amyloidosis may develop with end-stage renal disease as a consequence. In 1828 Marjolin described carcinomatous changes in burn scars.⁶⁵ In today's terminology, a *Marjolin ulcer* denotes malignant degeneration of chronic wounds, most commonly into squamous cell carcinoma. Unlike other epithelial tumors of this type, Marjolin ulcers tend to be more aggressive with early metastases and a morbid clinical course. In one series, 65% of patients developed metastases within 3 years of diagnosis, with a 2-year survival rate ranging from 66% to 80%. Prevention of systemic diseases, including amyloidosis and wound-related malignancies, is another reason to prevent chronic wounds in children.

FUTURE TREATMENT MODALITIES

Cytokine therapies have received attention in the wound-healing literature in recent years. Topical application of these agents, particularly platelet-derived growth factor (PDGF) (as recombinant human [rh]-PDGF-BB or becaplermin gel), has been studied extensively. Several randomized, double-blinded clinical trials have demonstrated efficacy of this agent as independent or adjunct therapy in healing chronic wounds.⁶⁶⁻⁶⁸ Kallianinen et al⁶⁶ showed that 92% of wounds pre-treated with PDGF remained closed after “salvage” surgery, compared with 0% of placebo-treated wounds in control subjects. Once-daily application of this mitogenic cytokine, at a concentration of 100 µg/g, not only significantly increased the incidence of complete wound closure (at least 90%) in another phase II randomized study but also significantly reduced the median relative ulcer volume, compared with placebo-gel counterparts.⁶⁷ Despite this evidence, PDGF-based therapies are not commonly used in many centers because of their high cost. Other emerging technologies in tissue engineering (for example, cultured keratinocytes) and molecular biology will probably contribute to improved outcomes in the management of pressure sores.

KEY POINTS

- Adult and pediatric pressure sores have in common unrelieved pressure.
- Children develop pressure sores in the same manner as adults but with some different anatomic distributions.
- Paramount to the successful nonoperative or operative treatment of pressure sores is patient education coupled with a multidisciplinary approach.
- Prevention is the most effective approach to pressure sore management.
- Ideal candidates for flap treatment are motivated, have a clean wound that has failed to close despite appropriate nonoperative management, and understand their role in wound care and long-term pressure relief.
- Successful flap treatment of pressure sores includes adequate debridement of inflamed and thickened tissue, removal or reduction of bony prominences, and full flap mobilization on the pedicle for a tension-free wound closure.
- The postoperative management of pressure sores determines long-term outcomes and recurrence rates.
- Evidence shows that surgical treatment of pressure sores in children leads to lower recurrence rates than in adults and may be preferable to nonoperative management.

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65

Vascularized Composite Allotransplantation in the Pediatric Population

Siba Haykal • Ronald M. Zuker



Despite substantial advances in reconstructive surgery, many pediatric conditions do not have acceptable solutions. These include significant facial deformities (congenital and acquired) that cannot be satisfactorily reconstructed, even with current advanced techniques. Abdominal wall deficiencies may limit potential small bowel allotransplantation. Reconstruction of extensive anomalies and traumatic loss of the extremities is particularly challenging. Current techniques often fall short of successful reconstruction for patients with laryngeal defects, uterine anomalies negating the possibility of childbirth, and micropenis.

In this chapter, we will present a brief history of vascularized composite allotransplantation (VCA) and attempt to place this new surgical tool in perspective. We will discuss the scientific basis of VCA and the research performed to date. The chapter will focus on some of the specific issues that apply to children. What are the effects on growth and function of the allograft? How does immunosuppression affect a growing child? What are the ethical concerns regarding pediatric transplantation? Last, we will explore the potential indications for pediatric VCA.

HISTORICAL PERSPECTIVE

Sushruta,¹ the founding father of surgery, described the surgical transfer of tissue from one location to another. In the Sushruta Samhita—Sushruta's compendium, written around 600 BC—he described the process of nasal reconstruction. The nose was a symbol of dignity and respect in ancient India and was frequently amputated in those who committed criminal offenses, adulterers,

Box 65-1 Sushruta's Original Sanskrit Description of Nasal Reconstruction

"The portion of the nose to be covered should be first measured with a leaf. Then a piece of skin of the required size should be dissected from the living skin of the cheek and sutured back to cover the nose, keeping a small pedicle attached to the cheek. The part of the nose to which the skin is to be attached should be made raw by cutting the nasal stump with a knife. The physician should then place the skin on the nose and stitch the two parts swiftly, keeping the skin properly elevated by inserting two tubes of erand (the castor oil plant) in the position of the nostrils so that the new nose gets proper shape. The skin thus properly adjusted, should then be sprinkled with a powder of licorice, red sandalwood and barberry plant. Finally it should be covered with cotton, and clean sesame oil should be constantly applied. When the skin has united and granulated, if the nose is too short or too long, the middle of the flap should be divided and an endeavor made to enlarge or shorten it."

From Saraf S, Parihar R. Sushruta: the first plastic surgeon in 600 B.C. Internet J Plast Surg 4, 2006.



Fig. 65-1 Sushruta—the founding father of surgery.

and prisoners of war. Tissue was transferred from the cheek to the nose, and Sushruta's description from two and a half centuries ago reads like a modern operative note, including the description of a template for the defect and a complex postoperative dressing (Box 65-1 and Fig. 65-1).

This procedure was modified, probably by one of his many students, as a transfer of an arterialized flap from the forehead—the classic Indian rhinoplasty¹ (Fig. 65-2). After generations, the concept and description of this procedure were brought to Europe, and distant transfers evolved, as described by the pioneers of reconstructive surgery Tagliacozzi and Curtorum² (Fig. 65-3). They said, "We restore, rebuild, and make whole those parts which nature hath given, but which fortune has taken away. Not so much that it may delight the eye, but that it might buoy up the spirit and help the mind of the afflicted."

The "first" recorded case of VCA is thought to be the legendary story of Saints Cosmas and Damian, twin brothers and physicians. Their famous, miraculous exploit was the grafting of a

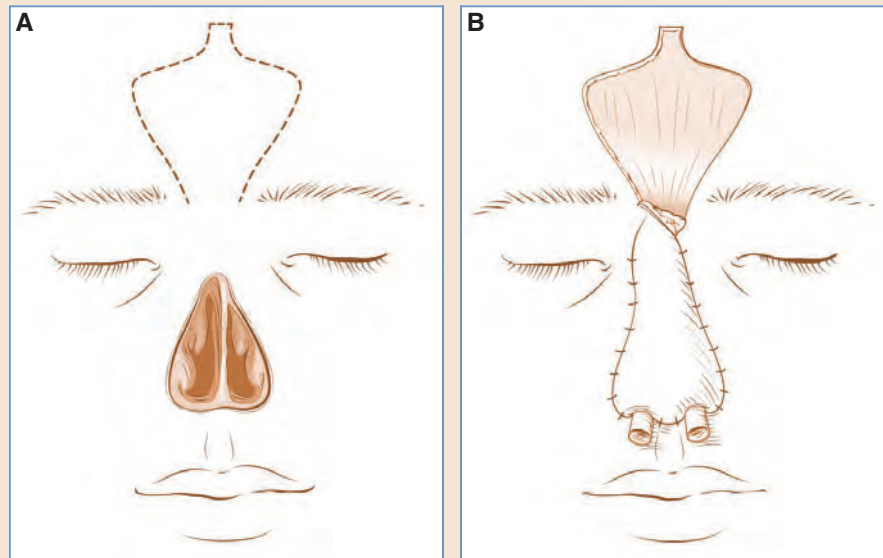


Fig. 65-2 Modification of rhinoplasty using an arterialized forehead flap—the classic Indian rhinoplasty—instead of a cheek flap. **A**, Preoperative markings. **B**, The flap is transferred into position.



Fig. 65-3 The original drawing of a so-called Italian method of rhinoplasty. The Indian surgeons of antiquity performed this distant transfer of vascularized tissue, which reconstructive surgeons of the twentieth century expanded into a free vascularized tissue transfer.



Fig. 65-4 Depiction of Saints Cosmas and Damian by artist Ambrosius Francken the Elder in the late sixteenth century. They are said to have carried out the first recorded VCA by transferring tissue from one person to another.

leg from a recently deceased Ethiopian to replace their patient's ulcerated leg³ (Fig. 65-4). The outcome of the surgery is unknown, but the Christian twins and their younger, collaborating brother were put to death by the Roman authorities for heresy.

The work of Billingham et al⁴ on World War II burn victims led to the concepts of graft rejection and acquired immune tolerance. A badly burned patient who had received skin grafts from the same donor on two different occasions rejected the second graft more rapidly than the first graft, suggesting immunologic memory. Rejection could be adoptively transferred in animals if they received the sensitized blood cells but not serum of the original graft recipient. These studies laid the foundation of immune tolerance and gave hope to patients and researchers that the immune system might be manipulated to develop tolerance to foreign tissue in the complete absence of drug-induced immunosuppression.

Murray,⁵ an American plastic surgeon who won the Nobel Prize for his work in organ transplantation, promoted the involvement of plastic surgeons in the field of transplantation. He emphasized the importance of balancing the immunologic and physiologic factors of organ transplantation and proposed a relative scale of antigenicity of tissues and organs. The immunologic factors included donor antigenicity, host responsiveness, genetic compatibility, and the immunosuppressive regimen. The physiologic factors were the function of the donor kidney, the preservation method, the ischemic interval, the anastomoses of vessels and the ureter, the lung-liver-reticuloendothelial function, and infection. According to his relative scale of antigenicity of tissues and organs, the skin and lungs are the most antigenic, the liver and heart are less antigenic, and the kidneys and pancreas are the least antigenic.⁵ Murray recognized the

Fig. 65-5 Matthew Scott received a new hand in 1999 in Louisville, Kentucky. It was the first successfully functioning hand transplant.



importance of identical twins having identical immunity and successfully carried out the first kidney transplant.⁶ He later used radiation to reduce rejection and introduced immunosuppressive drugs to control rejection, ushering in the new world of organ transplantation.

In 1963 the first hand transplantation was performed in Ecuador, but it survived for less than 3 weeks because of ineffective immunosuppressive therapy.^{7,8} Advances in immunosuppression therapy led to more innovations. A human hand allograft was performed in Lyon, France, in 1998 but was rejected and removed 29 months postoperatively, because the patient did not comply with the immunosuppressive treatment.^{9,10} A team of plastic and orthopedic surgeons led by Breidenbach^{11,12} and Tsai performed the first successful hand transplant in Louisville, Kentucky. The patient was carefully selected on the basis of anatomic need, rehabilitation potential, psychosocial stability, and assurance of maintenance of immunotherapy. The result was superb, far beyond what could be achieved with a prosthesis (Fig. 65-5). However, this functional success had a price. With immunosuppressive therapy, one of the Louisville patients developed diabetes, and another developed avascular necrosis of the hip.¹² In 2005 a partial face was successfully transplanted in a 38-year-old woman.¹³ The result was remarkably far better than could be achieved with current reconstructive surgery techniques (Fig. 65-6). Since then, more than 28 facial transplants¹⁴ and more than 100 upper extremity transplants¹⁵ have been performed around the world. In 2012 a 19-year-old boy received a total face transplant in Turkey.¹⁶ He is the youngest successful face transplant recipient to date.

In 2015 the first pediatric VCA was performed in an 8-year-old boy with quadrimembral amputations from sepsis. The Philadelphia team, led by Scott Levin (Fig. 65-7; see Fig. 65-12), carried out the surgery. The early results are promising and may set the standard for pediatric extremity VCA in the future. We have learned from the current experience with adult patients that these VCA procedures can achieve results not possible by any other means. Critical factors in patient selection include anatomic need, rehabilitative potential, psychosocial stability, and solid support systems. Surgeons should be confident that patients can comply with the immunosuppressive regimen. The survival of the patient, the survival of the allotransplant, and the function of the tissue components depend on these factors.



Fig. 65-6 Isabelle Dinoire received the first face transplant in 2005. She has a remarkable aesthetic and functional result.

Fig. 65-7 Dr. Scott Levin led the team that performed the first successful VCA in a child. He is seen with his young patient in the early postoperative period. (Courtesy of Children's Hospital of Philadelphia.)



Other VCA tissues have been transplanted, with results varying from excellent (as in the Scandinavian experience with uterine transplants¹⁷) to complete failure (as in the Chinese experience with a penile transplant¹⁸).

THE SCIENCE OF VASCULARIZED COMPOSITE ALLOTRANSPLANTATION

VCAs are composed of many different tissues, including skin, fat, muscle, nerve, vessel, and bone. They differ from solid-organ allografts such as the heart, lung, and liver in many ways. Some are highly visible, such as face and hand allografts. They are not life saving, but rather life enhancing.¹⁹ Thus new issues for discussion arise regarding indications and ethics of an elective procedure, with significant, long-term complications revolving around lifelong immunosuppression. This is even more controversial in children, as discussed later in the chapter. In this section, we review the evolution of our understanding of the immunology associated with VCA and the cur-

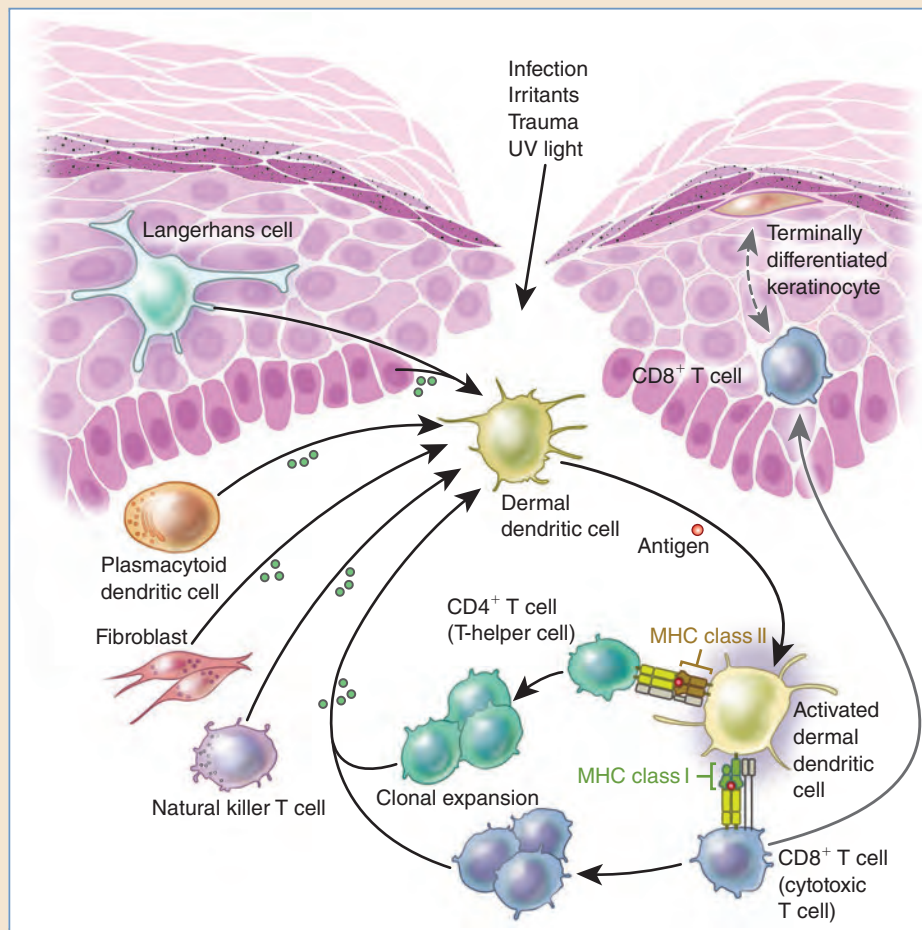


Fig. 65-8 The skin epidermis is considered a privileged target of graft rejection, because it contains numerous specialized APCs, such as Langerhans cells, and nonspecialized APCs, such as dendritic cells. (MHC, Major histocompatibility complex.)

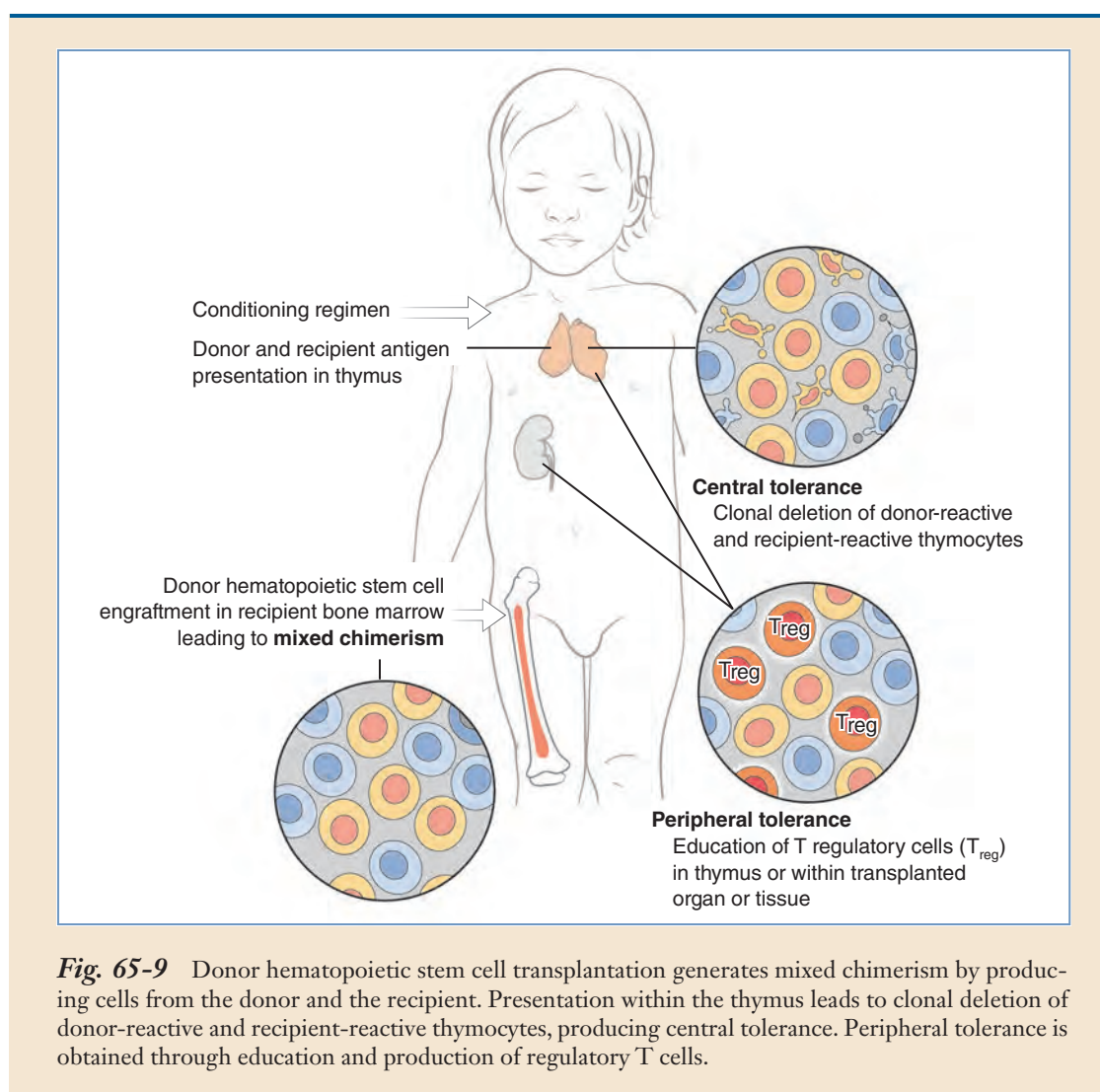
rent basic science research in this field. Presently, questions regarding immunology in VCA are the following:

- Is VCA feasible considering the high antigenicity of the skin component?
- Can VCA be performed without the use of highly toxic doses of immunosuppressive agents?
- Can VCA patients develop tolerance to reduce or eliminate immunosuppression?

In 1957 Billingham and Brent²⁰ discussed the concept of *split tolerance*. It refers to simultaneous tolerance to one tissue and rejection of another from the same donor. Skin grafts almost always fail before any other transplanted tissue or organ.²⁰ The secondary vascularization required for acceptance of conventional skin grafts upregulates cytokines, leading to rejection. The skin is highly immunogenic, because it contains a large number of specialized antigen-presenting cells (APCs), such as human leukocyte antigen class II APCs (Langerhans cells), and nonspecialized APCs, such as dendritic cells. Therefore the epidermis is considered a privileged target of graft rejection²¹ (Fig. 65-8).

In 1991 Lee et al²² published a key paper in the field of VCA. They transplanted donor rat limbs into rat recipients. The limbs were transplanted either as a whole or after division into different components (skin, subcutaneous tissue, muscle, bone, and blood vessel). They noted that different components caused different responses at a cellular or humoral level. Skin was not the most immunogenic. The limbs transplanted as a whole caused a lower response, compared with individual components. The authors hypothesized that this was the result of a “consumption” phenomenon and antigen competition. They also observed a higher immune response toward the vascularized limbs, compared with the nonvascularized limbs. This focused attention on split tolerance.

The ultimate goal is to induce tolerance. This can be achieved through preconditioning regimens that “prepare” the host to receive donor tissues and organs. Three main concepts emerge: macrochimerism, microchimerism, and mixed chimerism. In macrochimerism, the donor pluripotent hematopoietic stem cells engraft in the recipient and produce all its lineages, including the donor immune system; a new, hybrid immune system is established in the recipient and



reciprocal bidirectional donor, which leads to host tolerance.²³ In microchimerism, passenger leukocytes from the transplanted allograft interact with the recipient leukocytes and are hypothesized to lead to clonal exhaustion, resulting in donor-specific tolerance; this may explain how some transplant recipients require significantly reduced levels or no immunosuppression at all over time.²³ In mixed chimerism, the donor and recipient hematopoietic systems coexist. In humans, bone marrow transplantation–induced mixed chimerism has been shown to confer acceptance of donor-specific skin and kidney allografts without long-term immunosuppression. Mixed chimerism prevents chronic rejection, which is the major cause of late graft loss. This is probably associated with a lower incidence and severity of graft-versus-host disease, retains immunocompetence for primary immune responses, and can be induced through nonmyeloablative conditioning²⁴ (Fig. 65-9).

After conditioning is carried out to suppress alloreactivity and to facilitate engraftment, donor hematopoietic stem cell transplantation is performed. Engraftment of donor hematopoietic stem cells in recipient bone marrow results in persisting mixed chimerism and a self-renewing source of progeny for education in the thymus. Coexistence of donor and recipient antigen-presenting cells in the thymus results in clonal deletion of donor-reactive and recipient-reactive thymocytes (central tolerance), with tolerance of transplanted organs or tissues and a reduced risk of graft-versus-host disease. Presumably, alloreactive thymocytes escaping deletion are controlled by T regulatory cells (peripheral tolerance), educated either in the thymus or within transplanted tissues (see Fig. 65-9). Why is the immunogenicity of the whole limb less than that of the skin alone? One possibility is that VCAs (particularly limb transplants) function as a vascularized bone marrow transplant.^{25,26}

Animal and Preclinical Models of Vascularized Composite Allotransplantation

Several animal models have been developed to investigate the immune responses to different VCA components. Hettiaratchy et al²⁷ performed heterotopic limb transplantation across both haploidentical and fully mismatched major histocompatibility complex barriers after a nonmyeloablative, radiation-free conditioning protocol. Limb transplant and donor hematopoietic cell infusion with either bone marrow or cytokine-mobilized peripheral blood mononuclear cells (CMPBMC) were performed while animals received a 30-day course of cyclosporine A. Split tolerance was observed, and skin was rejected by day 60. Mixed chimerism was detected in recipients of CMPBMC but not in bone marrow recipients; however, animals from both groups developed tolerance of musculoskeletal tissues and in vitro evidence of donor specific unresponsiveness. Furthermore, all animals with demonstrable chimerism developed cutaneous graft-versus-host disease approximately 70 days after the transplant. All cases resolved with a short course of cyclosporine or methylprednisolone; however, this was a significant complication that probably would not be considered acceptable in a clinical setting.

VCAs are an ideal form of bone marrow transfer, because the graft itself provides the stromal microenvironment for proliferation of donor cells. However, studies in large animals failed to demonstrate lasting chimerism.²⁴

Because the graft is visible, biopsy samples are easily obtained at different stages but can often lead to bias. For example, Cendales et al²⁸ proposed the Banff grading system in which, despite the histologic evidence of infiltration, some changes do not require an increase in the immunosuppressive dose. They were able to assess the phenotype of infiltrating cells that led to the identification of facilitating and tolerogenic cells.²⁹

VCA appears to be feasible. As a whole organ in combination with other tissues, the skin does not behave the same as when it is transplanted alone. Advances in solid-organ transplantation, particularly immunosuppressive techniques, will help in preventing the use of highly toxic doses

in VCA. The preclinical large animal studies are promising with regards to tolerance, which remains transient in the clinical cases of VCA.

The current focus of research is on cell therapy, adhesion molecules, chemokines and cytokines, and tertiary lymphoid organs, with the ultimate goal of tolerance.

ADVANCEMENTS IN IMMUNOSUPPRESSION

The growth of solid-organ transplantation parallels the emergence of immunosuppressive drugs. In the 1960s azathioprine and prednisolone were used in renal transplantation.³⁰ Polyclonal antithymocyte globulin preparations became available in the 1970s.³⁰ The introduction of cyclosporine A in the early 1980s improved 3-year graft survival. Subsequently, the use of tacrolimus and sirolimus led to the current 1-year survival rate of 94% for kidney transplants, and monoclonal antibodies have led to steroid-sparing immunosuppression³⁰ (Box 65-2).

Currently, most immunosuppressive regimens include the following:

- Polyclonal antibodies: antithymocyte globulins or antiinterleukin 2 receptor antibodies (daclizumab and basiliximab)
- A triple-drug maintenance protocol consisting of a calcineurin inhibitor (tacrolimus), an antiproliferative agent (mycophenolate mofetil), and corticosteroids (prednisolone)³⁰ (Fig. 65-10)

The adverse effects and complications of these medications include the following:

- Opportunistic infections
- Malignancy
- Posttransplant lymphoproliferative disorders
- Hypertension
- Dyslipidemia
- Diabetes mellitus
- Renal insufficiency

However, without adequate immunosuppression, the risk of developing chronic rejection and graft loss is almost certain.

Pediatric solid-organ allograft survival rates have improved over the past years because of advances in immunotherapy regimens. Despite such advances, the failure rate is 25% to 35%.³¹ Many patients will require a second transplant, because transplantation rates for chronic rejection range from 5% to 25% for all organ types.³² Long-term immunosuppressive therapy exposes patients to an increased lifetime risk of infections, diabetes, hypertension, dyslipidemia, cardiovascular disease, renal insufficiency, and malignancy. Nonadherence to medication is a common cause of graft dysfunction and loss among the adolescent transplant recipients. Rejection episodes, hospitalizations, and medication adverse effects not only contribute negatively to the quality of life of the patients but can also affect growth and development and psychosocial stability.³³

Box 65-2 Five Broad Categories of Immunosuppressants

1. Glucocorticoids
2. Cytostatic agents
3. Antibodies
4. Drugs acting on immunophilins
5. Other drugs

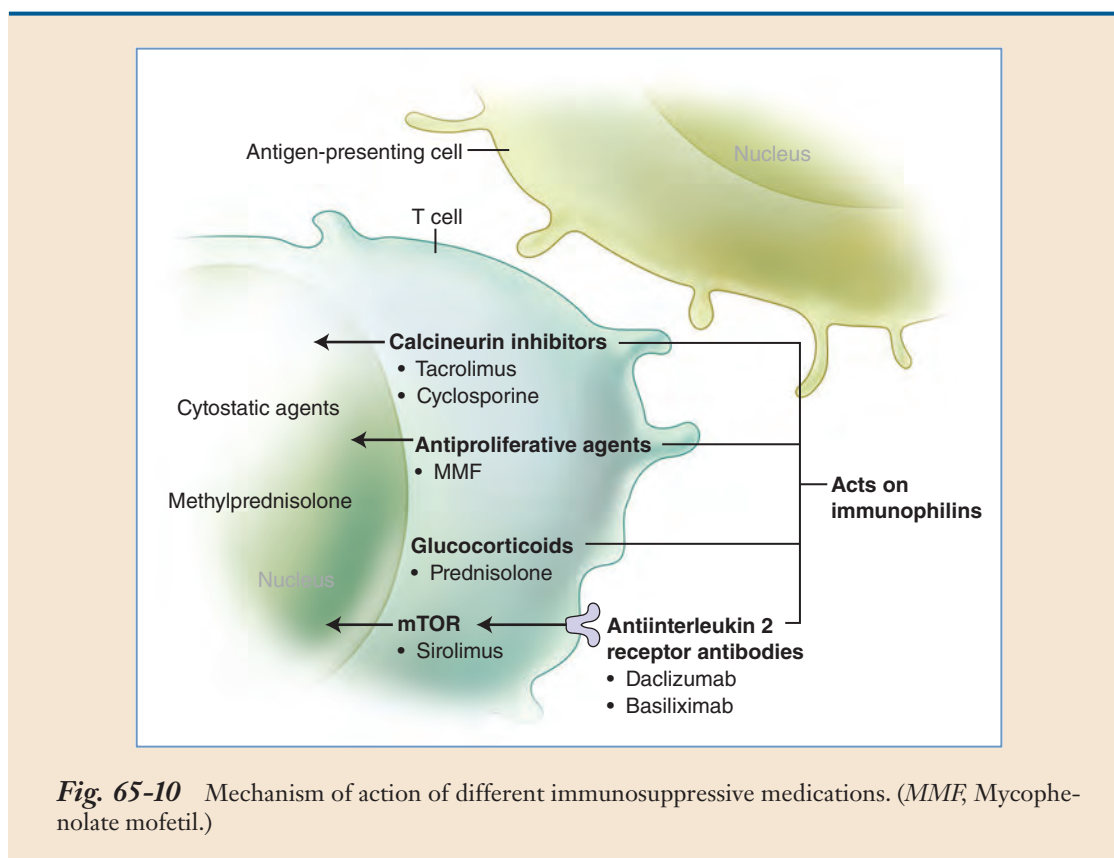


Fig. 65-10 Mechanism of action of different immunosuppressive medications. (MMF, Mycophenolate mofetil.)

The first reports on suspected chronic rejection in hand transplantation are now emerging, indicating amputation of the transplanted limb.³⁴ Chronic rejection in face transplant recipients has not been reported. (The longest follow-up is 8 years.) The effects of such a complication could prove to be devastating and potentially fatal.

SPECIFIC ISSUES FOR VASCULARIZED COMPOSITE ALLOTRANSPLANTATION IN CHILDREN

Much of what we anticipate for VCA in children is predicated on our experience in replantation and free tissue transfer, especially regarding growth and function. Similarly, the effects of immunosuppression in children are based on our solid-organ transplant experience. Several unknowns exist in the field of ethics, about which we can only hypothesize. Answers to many of these ethical concerns will evolve. This section will discuss the specific issues of VCA in children and conclude with a list of complex, insolvable problems for which VCA may be indicated.

Growth and Function of Allograft

Good evidence from our replantation experience shows that the growth of the replanted part will continue in an uninterrupted fashion if the growth plate is uninjured and has undergone only minimal ischemia. The growth plate is known to be quite sensitive to ischemia; therefore replantation in a timely fashion has always been critical in growing children. We anticipate that VCA



Fig. 65-11 Long bone growth in a transplant may be nearly normal if both the metaphysis and the epiphysis are revascularized. This radiograph shows bony union and maintenance of structure.

will have a similar pattern. In free tissue transfer of growth plates from one part of the body to another, such as in a toe transfer or a fibular transfer, growth will continue as it would have in the original location of the transplant. Tissues will grow at a programmed rate. This emphasizes the importance of transplanting like to like tissue of compatible age that is projected to have similar height and weight as the recipient.

Bowen et al³⁵ conducted research showing that long bone growth is “near normal” or “about 90% of normal” if both the metaphysis and the epiphysis are revascularized (Fig. 65-11). These studies were conducted years ago and have been reconfirmed. Therefore we agree that transplants will grow at near-normal rates.

The next question is, “Will the transplant function?” The International Registry on Hand and Composite Tissue Transplantation contains cumulative data regarding the function of hand transplants.¹⁰ More than 10 years of outcome data is recorded. The data for initial function and long-term improvement are very positive. The Registry has shown that the Disability of the Arm, Shoulder, and Hand scores decrease over time, indicating improved function and reduced disability. Similarly, the Hand Transplantation Score System improved initially and continued to improve steadily during the 10-year period¹⁰ (Table 65-1). This indicates ongoing improvement in hand function after the transplant. Based on the results, we think that where a part was present at birth and has been lost from trauma or disease, function can be regained through VCA.

Where There Was Cortical Representation at Birth In some pediatric conditions, a part is not present at birth. The usefulness of VCA in these patients is questionable. However, we think that where cortical representation is present, for example, in an intrauterine amputation with amniotic band syndrome, this cortical component will reform readily with a transplant. The cortical representation that was present previously should return to its previous state after a VCA, and appropriate cortical representation and near-normal integrative function should develop. Lundborg³⁶ documented this phenomenon, which is thought to be particularly useful in younger patients.

Where There Has Never Been Cortical Representation It is not known whether a child born without an anatomic part and with no cortical representation of that part will acquire an integrated activity through the process of cortical plasticity after a VCA. There is good evidence that cortical plasticity will provide integrative function. This has been shown in facial paralysis reconstruction in which the trigeminal nerve is used to reconstruct the seventh nerve.^{37,38} It has also been demonstrated in brachial plexus reconstruction in which the intercostal nerves are used

Table 65-1 Hand Transplantation Score System for Bilateral Hand Transplants

Date of Transplantation Follow-up (yrs)	2000		2000		2003		2003		2006	
	7		7		4		4		1	
	R	L	R	L	R	L	R	L	R	L
Appearance (0-15)	12	12	13.5	13.5	14	14	12.5	12.5	12.5	12.5
Sensibility (0-20)	18.5	19	17	17	16	19	9	10	12	12
Movement (0-20)	10.5	10.5	20	19.5	15	16.5	14.5	15	14	13.5
Psychological and social acceptance (0-15)	15	15	14	14	15	15	13	12	14	14
Daily activities and work status (0-15)	15	13	15	15	7	7	8	5	7	6
Patient satisfaction and general well-being (0-15)	15	15	15	15	11	11	6	6	6	6
TOTAL	86	84.5	95	94	78	82.5	63.5	60.5	65.5	64

L, Left hand; R, right hand.

The Hand Transplantation Score System evaluates six aspects with different weight for a total of 100 points: appearance (15 points), sensibility (20 points), movement (20 points), psychological and social acceptance (15 points), daily activities and work status (15 points), and patient satisfaction and general well-being (15 points). A total result of 81 to 100 points is graded as an excellent outcome, 61 to 80 as good, 31 to 60 as fair, and 0 to 30 as poor.

to power elbow flexion.³⁹ In a case of conjoined twins, one of whom survived, the surviving twin underwent a lower extremity transplant of her twin sister's leg. She had no cortical representation of the leg preoperatively. Because of cortical plasticity, she developed integrative function after the transplant. This was documented clinically and by MRI.³⁹⁻⁴¹

Thus, although we do not know for sure, we anticipate that with the aid of cortical plasticity, children who are born without parts and never had cortical representation may be suitable candidates for a VCA. Thus the lack of cortical representation of a missing part at birth should not necessarily preclude the possibility of transplantation and a worthwhile functional outcome.

The Effect of Immunosuppression in Children

Growth and Development

Children who receive solid-organ transplants must continue immunosuppressive therapy for life. Several studies have shown that immunosuppression does not impair growth.^{42,43} Many patients with end-stage renal disease have impaired growth potential and undergo catch-up growth after a kidney transplantation.^{42,43} Similarly, in liver transplantation patients, growth does not seem to be impaired with the use of long-term immunosuppressants.

Assessing growth and development can be difficult in patients with chronic disease. The studies on heart transplants in infants may provide better evidence. Studies by Hirsch et al⁴³ at Washington University and by Chinnock et al⁴⁴ at Loma Linda University showed normal growth and development in these children, despite immunosuppressive therapy.

The current thought is that growth and development alteration after a pediatric organ transplantation are related to long-term corticosteroid use.⁴⁵ Current efforts are aimed at reducing steroid use to low-dose levels, alternate-day dosing, or even to none at all. Reducing rejection episodes on this basis is critical, because steroids are often reintroduced or dosing increased to combat such episodes.

Lymphoproliferative Disease

Immunosuppressive therapy does not seem to alter growth or development in children. However, does the incidence of lymphoproliferative disease increase over time? The renal transplant data on this topic are excellent, specifically data from the North American Pediatric Renal Trials and Collaborative Studies.⁴⁶ In more than 10,000 transplants, the incidence of lymphoproliferative disease was 2.33%, and most occurred after 10 years. This relatively low rate is anticipated to become even lower with the newer immunosuppressive drugs. However, the incidence varies with the type of organ transplanted; kidney transplants have the lowest incidence, and intestinal transplants have the highest.⁴⁷ Young children may be more susceptible, because they may not have not been exposed to the Epstein-Barr virus; they are five times more likely to develop post-transplant lymphoproliferative disease.⁴⁷ Thus this disease is a concern in young children with long-term immunosuppression, particularly those who are Epstein-Barr virus seronegative, emphasizing the need for a broad-view risk-benefit analysis.

Malignancy

Long-term immunosuppression can increase the risk for other malignancies, including skin cancers in children, particularly malignant melanoma and the virally mediated Kaposi sarcoma.⁴⁷

Hypertension

Hypertension is a common sequela to immunosuppression therapy in children.^{48,49} It not only increases the risk for cardiovascular disease, but can also contribute to renal dysfunction. The causes are varied and seem to be dependent on the agents given, emphasizing the need to minimize immunosuppression (particularly corticosteroid).⁵⁰

Metabolic Alterations

As in adults, many children taking long-term immunosuppressants develop dyslipidemia and diabetes mellitus.⁴⁹ Both can increase the risk for cardiovascular disease and have been implicated in allograft vasculopathy.^{50,51}

Renal Failure

Even in nonrenal transplant recipients, renal function can decline with long-term immunosuppression.^{50,51} This ranges from mild nephrotoxicity to total loss of renal function, necessitating dialysis or renal transplant.^{50,51} The causes are multifactorial, including immunosuppressive drugs (especially tacrolimus), nephrotoxic antibiotics, hypertension, and poor allograft function.^{50,51}

Infection

Infection is the leading cause of mortality in pediatric transplant recipients.^{52,53} It can originate in the community, the hospital, and the donor organ, or it may be a reactivation of a previous viral infection. The risk is greatest immediately after transplantation, when immunosuppression is greatest to reduce the likelihood of acute rejection. This risk decreases over time but is a lifelong concern. Infants and young children are at an even greater risk for infection. They have a naive immune system that may not have developed antibodies to certain microbes, they may not have completed their vaccination series, and they may not have been exposed to some viruses such as cytomegalovirus, Epstein-Barr virus, and herpes virus. Transplant recipients should be appropriately immunized, informed of the importance of avoiding potentially infective situations, and able to begin broad-spectrum antimicrobials at the first sign of infection.

Immunosuppression is a necessary evil in the field of transplantation. However, the following steps can be taken to minimize its adverse effects:

- Avoiding or treating infection early
- Reducing or eliminating long-term corticosteroid use
- Minimizing the immunosuppressive drug regimen
- Carefully monitoring for the potential malignant and metabolic consequences

Ethical Concerns in Children

Although VCA is technically feasible, the ethical issues are considerable, particularly in pediatric patients.^{16,54,55} Several major factors require consideration.

Risk-Benefit Analysis

The first concern is a risk-benefit analysis. Hand and facial transplantation are not lifesaving but rather offer a chance to improve lifestyle. Therefore, a patient's quality of life benefit should be evaluated. A recent study showed that facial transplantation can add up to 20 quality-adjusted life-years.⁵⁶ Hand transplantation in adults has been shown to lead to functional improvement.⁹ The patients and families must be able to endure the surgery, the postoperative rehabilitation, and the long-term, lifelong immunosuppression. This becomes more complex when the consent process involves a substitute decision-maker. Parents are often conflicted in their desire to help their child and evaluate the risk-benefit ratio. Because the future of VCA in children is unknown, this process is even more complex. Will the allograft deteriorate with time and need to be repeated? What are the implications of this for face transplant patients, whose identity will be altered yet again? We know from solid-organ transplantation that compliance in adolescence is often a problem. Will this be the case in VCA patients, and will allografts be lost because of noncompliance by teenage recipients?

Long-term complications of immunosuppression are unknown. Will children develop kidney failure or skin malignancies, which are known to be much more aggressive in the immunosuppressed population. In evaluating the risks versus benefits, we must be very open-minded about these unknowns. As noted previously, the growth of a transplant may not be normal, its ultimate fate is uncertain, and an acceptable exit strategy may not be available should complications arise.⁵⁷ In children, issues of personal identity and social integration are highlighted and compounded because of the lifelong risk of immunosuppression therapy for a nonlifesaving procedure.

Informed Consent

The second ethical issue is informed consent. In some jurisdictions that have set no age of consent, decisions regarding the ability of a child to act autonomously are made on a case per case basis. In many cases, the parents make the decision, which could potentially lead to future resentment from their children. The issue of informed consent is also relevant to donors and their families. Generally, adult patients and parents consenting to donate might appreciate assurance that the organs will be used appropriately. The outcomes of VCA in children are uncertain, and such an assurance cannot be provided. Furthermore, donating a child's face involves an emotional component, and grieving parents might find this especially difficult.

Personal Identity

Another ethical issue to consider is personal identity. This pertains in particular to face transplantation in pediatric patients. A new appearance might affect a child's psychological, mental,

emotional, and social well-being.⁵⁸⁻⁶³ This will influence the selection of candidates as they pass through development stages at different assessment times.⁶⁴⁻⁶⁸ The contribution of psychological health before and after trauma becomes very complex in this context.

Utility and Equity

Concerns regarding utility and equity need to be addressed in any discussion of ethical issues regarding VCA. The usefulness of the part and its benefit to the child must be clearly explained, including the risks, both short-term and long-term. A transplant is a most-valuable entity and must be offered to potential recipients in an equitable fashion. Proper processes must be in place to allocate this precious resource fairly and to those most in need.

INDICATIONS FOR PEDIATRIC VASCULARIZED COMPOSITE ALLOTRANSPLANTATION

Currently, we describe two broad categories of children who will undergo a VCA: (1) those who have begun or are about to begin immunosuppression to support a solid-organ transplant and (2) those who require immunosuppression for a VCA.

Patients Who Have Begun or Are About To Begin Immunosuppression To Support a Solid-Organ Transplant

For patients who have begun or are about to begin immunosuppression to support a solid-organ transplant, the risks of immunosuppression are not weighed, because it will be required for the solid-organ transplant anyway. The risk of the actual surgery, its consequences, and its benefits can be independently evaluated with much less attention given to the effects of immunosuppression. Some risks may be increased with an additional allogeneic load, but the long-term effects of the immunosuppressive regimen should be about the same as those of a solid-organ transplant alone.

For children already on a balanced, stable immunosuppressive regimen, this is the best scenario. The first pediatric VCA was recently carried out under these circumstances. Zion Harvey had a devastating infection as a toddler and lost all four limbs and the function of both kidneys. However, he survived and, after receiving a kidney transplant from his mother, lived a restricted life as a quadramembral amputee. He was treated by the team at the University of Pennsylvania in Philadelphia, which had previously treated a young woman who was a quadramembral amputee. She had a remarkably good outcome. They studied Zion carefully and concluded that he would be an excellent candidate for upper extremity transplantation. He was listed for transplantation, and the actual surgery was carried out in July 2015. The early follow-up shows successful revascularization (Fig. 65-12; see Fig. 65-7) and we await reports on the functional outcome.

This 8-year-old boy is the first child to undergo a VCA. A team of 40 carried out the surgery at The Children's Hospital of Philadelphia under the leadership of Dr. Scott Levin. This bilateral hand transplant will change this boy's life dramatically, with minimal added immunosuppressive burden, because he had previously undergone a kidney transplant. However, even with immunosuppression under way to support a solid-organ transplant, differences exist for the various types of VCA. An upper extremity transplant, for example, is readily reversible should it prove nonfunctional or undergo rejection for whatever reason. We do not know the longevity of a VCA, and this may be particularly relevant in a child. A nonfunctional or rejected upper extremity can be amputated and the pretransplant state nearly reestablished. The situation is quite different, however, for a face transplant or an abdominal wall transplant patient. Failure of the allograft and removal of the transplant would be very difficult, with significant morbidity, and potentially fatal.



Fig. 65-12 Zion Harvey is the first pediatric VCA recipient. At 8 years of age he underwent a double-hand transplant. The early reports are most encouraging. (Courtesy of Children's Hospital of Philadelphia.)

Immunosuppression Is Required Specifically for Vascularized Composite Allotransplantation

Although a VCA is not a lifesaving procedure, it can be enormously life enhancing. In this situation, in children who undergo immunosuppression specifically to support a VCA, the risks and long-term effects must be worthwhile. This is a delicate balance that requires weighing all of the issues discussed previously, such as growth and function of the allograft, the effect of immunosuppression in children, and the ethical concerns. Presently, such scenarios would be unusual, but nevertheless worthy of consideration. The scenarios can be categorized according to the type of VCA (Fig. 65-13).

Face Transplantation

Facial trauma is very common in children and can occur from physical injury, animal bites, and surgical intervention. Even with extensive trauma, our reconstructive techniques can produce excellent or acceptable results in most patients. Occasionally, however, it is impossible to achieve the desired results in children with severe midface injuries and extensive soft tissue or even bony damage. A VCA may be an option in these extensive central facial injuries.

Most congenital facial anomalies are reconstructible to an excellent or acceptable degree. However, some rare craniofacial defects involving the central part of the face are very difficult to reconstruct adequately, including some of the rare craniofacial clefts and the severe forms of Treacher Collins syndrome and hemifacial microsomia.⁶⁹ Consideration of a VCA is warranted for other rare central facial deficiencies such as arhinia and those that result from the destructive processes of massive midface hemangiomas, vascular malformations, and neurofibromatosis.

Tracheolaryngeal Transplantation

In patients with severe laryngeal clefts and loss of the stability of the laryngeal and tracheal rings, providing a secure, reliable airway and the anatomic structures required for voice production may not be possible. Laryngeal transplantation may play a role in these rare, selected cases of congenital stenosis, tracheobronchomalacia, and laryngeal clefts, where conventional reconstruction fails to provide what is needed.

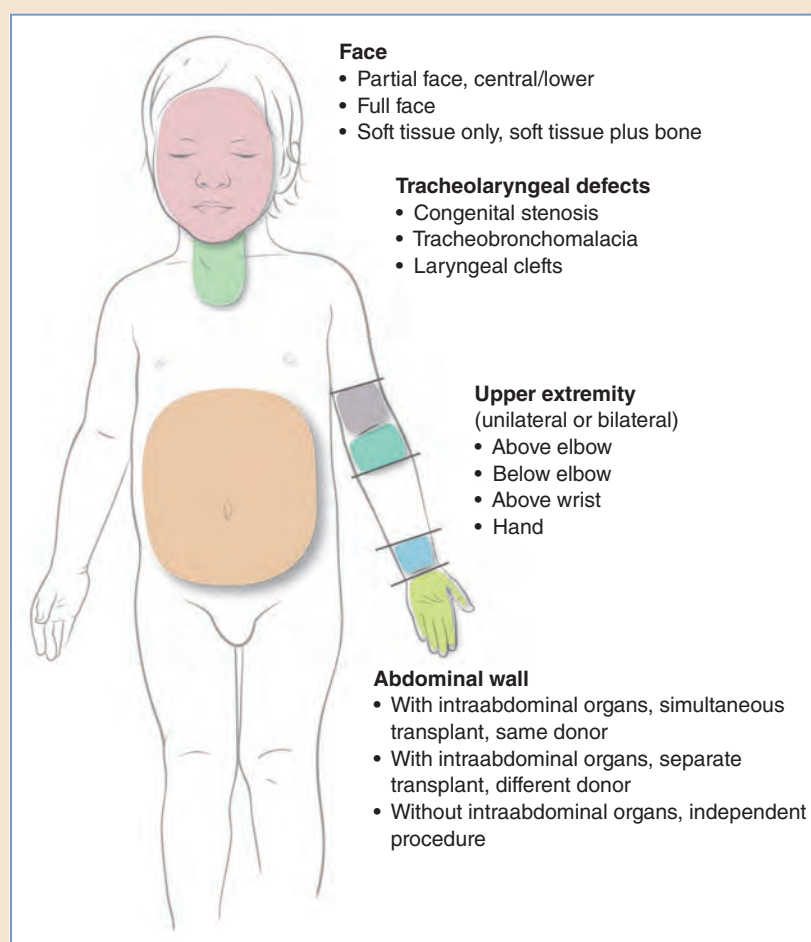


Fig. 65-13 Options for VCAs based on anatomic location.

Upper Extremity Transplantation

Children with congenital upper extremity anomalies function extremely well, compared with those who have similar, but acquired, anatomic deficiencies. Thus the indications for transplantation are more common in the group with acquired deficiencies. Traumatic injuries can result from trauma, surgery, or sepsis. Congenital anomalies that may be considered for a VCA include transverse arrest, amniotic band syndrome, and the extremes of overgrowth or vascular malformations. The factors to consider regarding suitability for a VCA include the functional impairment caused by the trauma or anomaly, bilaterality, the ineffectiveness of prosthetic use, and the overall benefit to a particular patient's quality of life.

Abdominal Wall Transplantation

Congenital anomalies of the gastrointestinal system can usually be reconstructed to an adequate degree. However, in rare circumstances the abdominal wall is absent, destroyed by an infective process or surgical intervention, and will not adequately support the abdominal organs. This can

occur with severe gastroschisis or with necrotizing enterocolitis. An abdominal wall transplant may be an option. This situation is most common in patients who also require an intraabdominal transplant.

THE FUTURE

VCA in children is an emerging field. Children who are already taking immunosuppressants and those who will require immunosuppressants for solid-organ transplantation may be candidates for a VCA for absent functionally significant anatomic parts. For children who do not take immunosuppressants, potential indications for a VCA include upper extremity loss, extensive mid-face and lower face deficits, abdominal wall deficiencies without adequate abdominal organ support, and extensive laryngotracheal deficiencies. These may be congenital or acquired through trauma, disease, or surgery.

VCA in general is an evolving area of surgical intervention. New indications are proposed, improved immunosuppressants are trialed, and a better understanding of infection control emerges. It is only a matter of time until these procedures are part of reconstructive surgeons' armamentarium for treating complex pediatric patients, providing opportunities for reconstruction never before possible.

KEY POINTS

- The history of VCA is a fascinating journey through medical history.
- The science of VCA is complex and becoming more complex with time.
- A transplant in children will grow and function in a manner similar to a replant or a free tissue transfer.
- Surgeons need to understand the effects of immunosuppression on growing children; in appropriate circumstances these should not be a barrier to a VCA.
- The ethical issues of risk-benefit ratios, informed consent for young children, personal identity in patients with visible transplants, utility, and equity need to be discussed openly, fully, and to the best of our knowledge.
- The indications for a VCA in children vary depending on whether immunosuppression has begun or is about to begin to support a solid-organ transplant and whether immunosuppression is required specifically to support the VCA.
- Options for potential VCA in children include the upper extremities, the face, the abdominal wall, and the tracheolaryngeal complex.

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